Evolutionarily Significant Biological Phenomena

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Mimicry

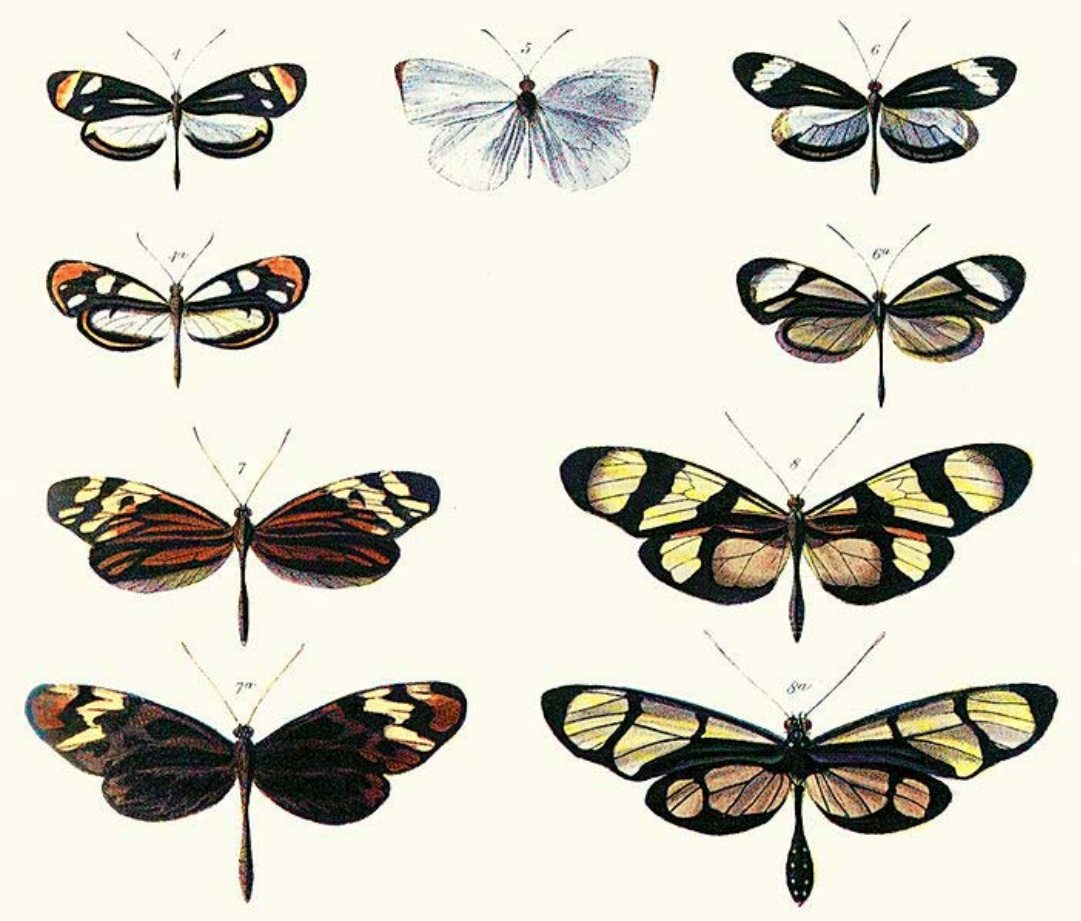


Plate from Henry Walter Bates (1862) illustrating Batesian mimicry between *Dismorphia* species (top row, third row) and various *Ithomiini* (Nymphalidae, second row, bottom row)

In evolutionary biology, **mimicry** is the similarity of one species to another which protects one or both. This similarity can be in appearance, behaviour, sound, scent and even location, with the mimics found in similar places to their models.

Mimicry occurs when a group of organisms, the *mimics*, evolve to share common perceived characteristics with another group, the *models*. The evolution is driven by the selective action of a *signal-receiver*, or *dupe*. For example, birds that use sight to identify palatable insects (the mimics), whilst avoiding the noxious models.

Collectively, this situation is known as a *mimicry complex*. The model is usually another species except in cases of automimicry. The signal-receiver is typically another intermediate organism like the common predator of two species, but may actually be the model itself, such as a moth resembling its spider predator. As an interaction, mimicry is in most cases advantageous to the mimic and harmful to the receiver, but may increase, reduce or have no effect on the fitness of the model depending on the situation. Models themselves are difficult to define in some cases, for example eye spots may not bear resemblance to any specific organism's eyes, and camouflage often cannot be attributed to a particular model.

A planthopper mimics a leaf (mimesis)

Camouflage, in which a species resembles its surroundings, is essentially a form of visual mimicry. In between camouflage and mimicry is **mimesis**, in which the mimic takes on the properties of a specific object or organism, but one to which the dupe is indifferent. The lack of a true distinction between the two phenomena can be seen in animals that

resemble twigs, bark, leaves or flowers, in that they are often classified as camouflaged (a plant constitutes its "surroundings"), but are sometimes classified as mimics (a plant is also an organism).[p51] Crypsis is a broader concept which encompasses all forms of avoiding detection, such as mimicry, camouflage, hiding etc.

Though visual mimicry is most obvious to humans, other senses such as olfaction (smell) or hearing may be involved, and more than one type of signal may be employed. Mimicry may involve morphology, behavior, and other properties. In any case, the signal always functions to deceive the receiver by preventing it from correctly identifying the mimic. In evolutionary terms, this phenomenon is a form of co-evolution usually involving an evolutionary arms race.[p161] It should not be confused with convergent evolution, which occurs when species come to resemble one another *independently* by adapting to similar lifestyles.

Mimics may have different models for different life cycle stages, or they may be polymorphic, with different individuals imitating different models. Models themselves may have more than one mimic, though frequency dependent selection favors mimicry where models outnumber mimics. Models tend to be relatively closely related organisms, but mimicry of vastly different species is also known. Most known mimics are insects, though many other animal mimics including mammals are known. Plants and fungi may also be mimics, though less research has been carried out in this area.

Etymology

Use of the word mimicry dates back to 1637. It is derived from the Greek term *mimetikos*, "imitative," in turn from *mimetos*, the verbal adjective of *mimeisthai*, "to imitate." Originally used to describe people, it was only applied to other forms of life after 1851.

Classification

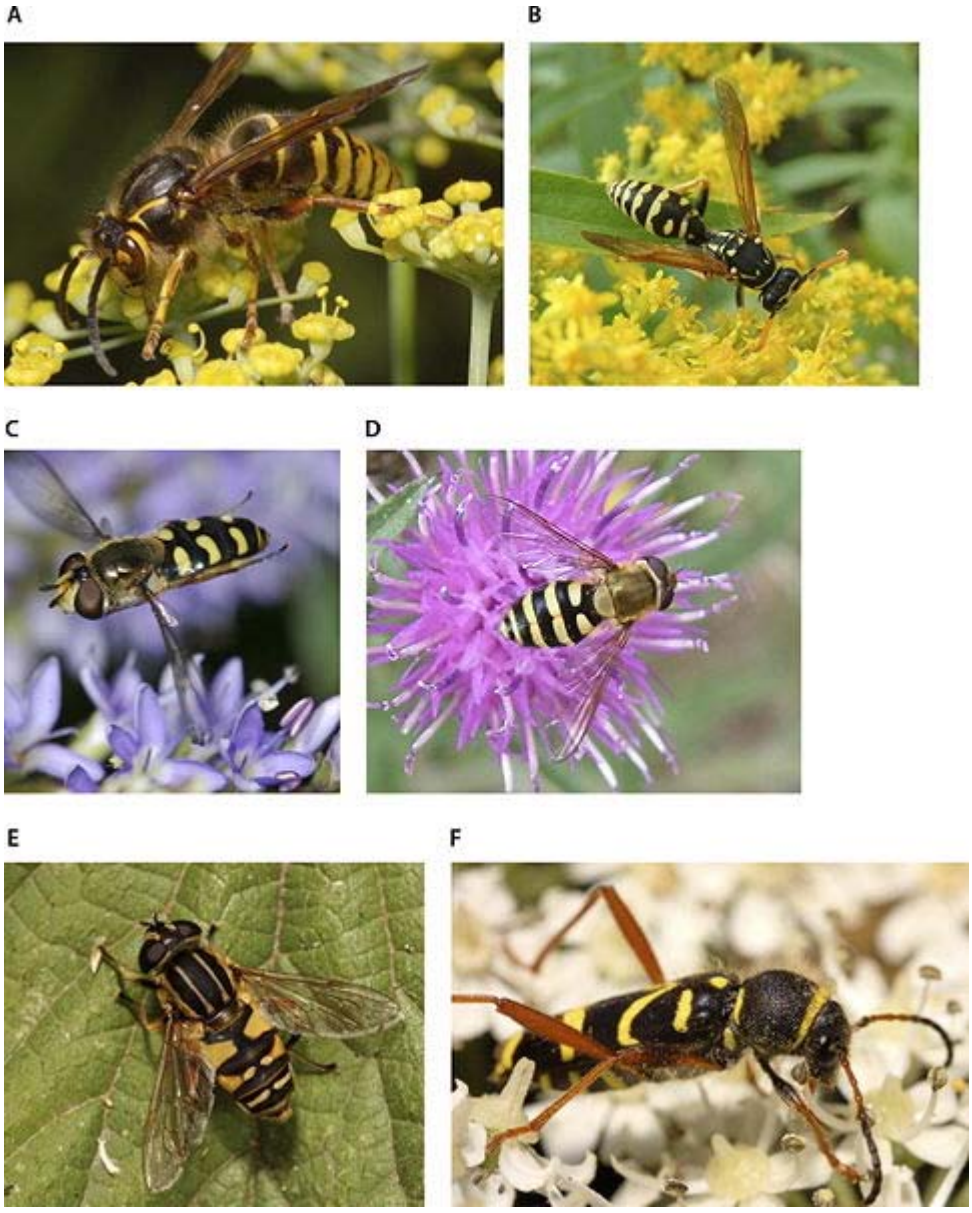
Many types of mimicry have been described. An overview of each follows, highlighting the similarities and differences between the various forms. Classification is often based on function with respect to the mimic (e.g. avoiding harm), though other parameters can also be used, and multidimensional classifications are required to understand the full picture. For this reason, some cases may belong to more than one class, e.g. automimicry and aggressive mimicry are not mutually exclusive, as one describes the species relationship between model and mimic, while the other describes the function for the mimic (obtaining food).

Defensive

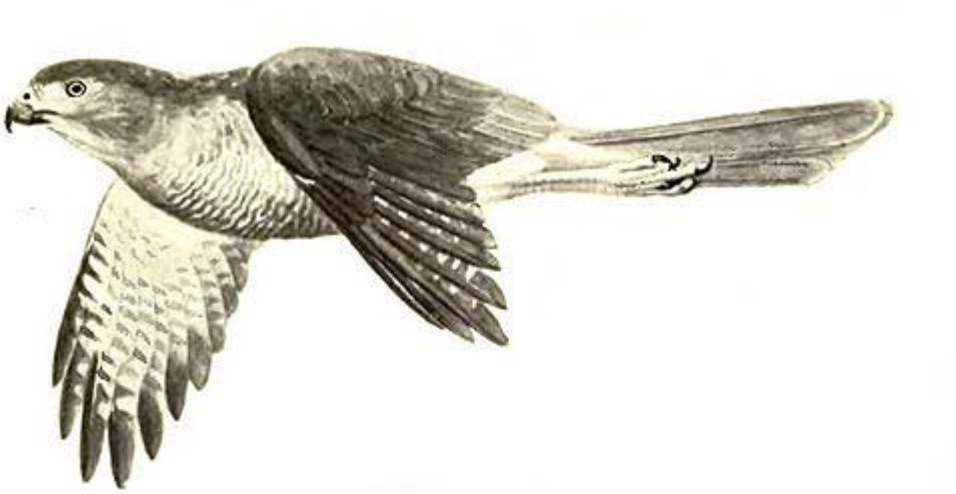
Macroxiphus sp katydid mimics an ant

Defensive or protective mimicry takes place when organisms are able to avoid an encounter that would be harmful to them by deceiving an enemy into treating them as something else. Four such cases are discussed here, the first three of which entail mimicry of an aposematic, harmful organism: Batesian mimicry, where a harmless mimic poses as harmful; Müllerian mimicry, where two harmful species share similar perceived characteristics; and Mertensian mimicry, where a deadly mimic resembles a less harmful but lesson-teaching model. Finally, Vavilovian mimicry, where weeds resemble crops, is discussed.

Batesian



Several species, including several hoverflies, mimic stinging species of wasp.



SHIKRA HAWK

The upper side of the tail is marked as in the Hawk-cuckoo

By permission of Messrs. Hutchinson & Co.

BRAIN-FEVER BIRD

The exact correspondence of this mimic with its model is notable even in black-and-white

By permission of Messrs. Hutchinson & Co.

Some hawk-cuckoos resemble hawks like the Shikra.

In Batesian mimicry the mimic shares signals similar to the model, but does not have the attribute that makes it unprofitable to predators (e.g. unpalatability). In other words, a Batesian mimic is a sheep in wolf's clothing. It is named after Henry Walter Bates, an English naturalist whose work on butterflies in the Amazon rainforest (including *Naturalist on the River Amazons*) was pioneering in this field of study. Mimics are less likely to be found out when in low proportion to their model, a phenomenon known as negative frequency dependent selection which applies in most other forms of mimicry as well. This is not the case in Müllerian mimicry however, which is described next. Examples:

- Lepidoptera

    - The Ash Borer (*Podosesia syringae*), a moth of the Clearwing family (Sesiidae), is a Batesian mimic of the Common wasp because it resembles the wasp, but is not capable of stinging. A predator that has learned to avoid the wasp would similarly avoid the Ash Borer.
    - Plain Tiger (*Danaus chrysippus*) – an unpalatable model with a number of mimics.
    - Common Crow (*Euploea core*) – an unpalatable model with a number of mimics.
    - *Consul fabius* and *Eresia eunice* imitate unpalatable *Heliconius* butterflies such as *H. ismenius*.
    - Several palatable butterflies resemble different species from the highly noxious papilionine genus *Battus*.
    - Several palatable moths produce ultrasonic click calls to mimic the unpalatable tiger moths.
- The False Cobra (*Malpolon moilensis*) is a mildly venomous but harmless colubrid snake which mimics the characteristic "hood" of an Indian cobra's threat display. The Eastern Hognose Snake (*Heterodon platirhinos*) similarly mimics the threat display of venomous snakes.
- The milk snake resembles the deadly coral snake.
- Vespid wasps bear several harmless mimics including moths, beetles and hoverflies.
- Octopuses of the genus *Thaumoctopus* (the Mimic Octopus) are able to intentionally alter their body shape and color so that they resemble dangerous sea snakes or lionfish.

Müllerian

The Heliconius butterflies from the tropics of the Western Hemisphere are the classical model for Müllerian mimicry.

Müllerian mimicry describes a situation where two or more species have very similar warning or aposematic signals and both share genuine anti-predation attributes (e.g. being unpalatable). At first Bates could not explain why this should be so; if both were harmful why did one need to mimic another? The German naturalist Fritz Müller put forward the first explanation for this phenomenon: If two species were confused with one another by a common predator, individuals in both would be more likely to survive. This type of mimicry is unique in several respects. Firstly, both the mimic and the model benefit from the interaction, which could thus be classified as mutualism in this respect. The signal receiver is also advantaged by this system, despite being deceived regarding species identity, as it avoids potentially harmful encounters. The usually clear identity of mimic

and model are also blurred. In cases where one species is scarce and another abundant, the rare species can be said to be the mimic. When both are present in similar numbers however it is more realistic to speak of each as *comimics* than of a distinct 'mimic' and 'model' species, as their warning signals tend to converge toward something intermediate between the two. Another theoretical problem comes up when one considers that the two species may exist on a continuum from the harmless to the highly noxious, raising the question of where Batesian mimicry ends and Müllerian convergence begins.

Examples:

- Lepidoptera
  - The Monarch Butterfly (*Danaus plexippus*) is a member of a Müllerian complex with the Viceroy butterfly (*Limenitis archippus*) in shared coloration patterns and display behavior. The Viceroy has subspecies with somewhat different coloration, each one very closely matching the local *Danaus* species. E.g., in Florida, the pairing is of the Viceroy and the Queen Butterfly, and in Mexico, the Viceroy resembles the Soldier Butterfly. Therefore, the Viceroy is a single species involved in three different Müllerian pairs. This example was long believed to be a case of Batesian mimicry, with the Viceroy being the mimic and the Monarch the model, but it was more recently determined that the Viceroy is actually the *more* unpalatable species, though there is considerable individual variation. While *L. archippus* is really bad-tasting, *Danaus* species tend to be toxic rather than just repugnant, due to their different food plants.
  - Unpalatable *Euploea* species look very similar.
  - The genus *Morpho* is palatable but are very strong fliers; birds – even species which are specialized for catching butterflies on the wing – find it very hard to catch them. The conspicuous blue coloration shared by most *Morpho* species seems to be a case of Müllerian mimicry.
  - The "orange complex" of species, including the heliconiines *Agraulis vanillae*, *Dryadula phaetusa*, and *Dryas iulia* which all taste bad.
  - Many different tiger moths make ultrasonic clicking calls to warn bats that they are unpalatable. Presumably a bat may learn to avoid *any* signalling moths, which would make this an example of Müllerian mimicry.
- Various bees and numerous vespid and sphecoid wasps: These animals are examples of Müllerian mimics because they have the aposematic yellow and black stripes (sometimes black and red, or black and white). Females of most of these species are potentially harmful to predators, fulfilling the second requirement of Müllerian mimicry. However, in essentially all such species, the males are harmless, and can thus be considered automimics of their conspecific females (see below). There are also many genera in these groups where the females are not capable of stinging, and yet still possess aposematic coloration (e.g., the wasp genus *Cerceris*), so they are considered Batesian mimics.

Emsleyan/Mertensian

Texas Coral Snake, *Micrurus tener* (left) and Mexican Milk Snake, *Lampropeltis triangulum annulata* (right).

Emsleyan or *Mertensian mimicry* describes unusual cases where deadly prey mimic a less dangerous species. It was first proposed by Emsley as a possible answer for the problem of Coral Snake mimicry in the New World. It was elaborated on by the German biologist Wolfgang Wickler in a chapter of *Mimicry in Plants and Animals*, who named it after the German herpetologist Robert Mertens. Sheppard points out that Hecht and Marien put forward a similar hypothesis ten years earlier.

This scenario is a little more difficult to understand, as in other types of mimicry it is usually the most harmful species that is the model. But if a predator dies, it cannot learn to recognize a warning signal, e.g. bright colors in a certain pattern. In other words, there is no advantage in being aposematic for an organism that is likely to kill any predator it succeeds in poisoning; such an animal would rather profit from being camouflaged, to avoid attacks altogether. If, however, there is some other species that is harmful but *not* deadly as well as aposematic, the predator may learn to recognize its particular warning colors and avoid such animals. A deadly species will then profit by mimicking the less dangerous aposematic organism, if this results in less attacks than camouflage would.

The exception here, ignoring any chance of animals learning by watching a conspecific die, is the possibility of not having to learn that it is harmful in the first place: instinctive genetic programming to be wary of certain signals. In this case, other organisms could benefit from this programming, and Batesian or Müllerian mimics of it could potentially evolve. In fact, it has been shown that some species do have an innate recognition of certain aposematic warnings. Hand-reared Turquoise-browed Motmots (*Eumomota superciliosa*), avian predators, instinctively avoid snakes with red and yellow rings. Other colors with the same pattern, and even red and yellow *stripes* with the same width as rings, were tolerated. However, models with red and yellow rings were feared, with the birds flying away and giving alarm calls in some cases. This provides one alternative explanation to Mertensian mimicry.

Examples:

- Some Milk Snake (*Lampropeltis triangulum*) subspecies (harmless), the moderately toxic False Coral Snakes (genus *Erythrolamprus*), and the deadly Coral Snakes all have a red background color with black and white/yellow rings.

In this system, both the milk snakes and the deadly coral snakes are mimics, whereas the false coral snakes are the model.

Wasmannian

Wasmannian mimicry refers to cases where the mimic resembles a model along with which it lives (inquiline) in a nest or colony. Most of the models here are social insects such as ants, termites, bees and wasps.

Mimetic weeds

Rye is a secondary crop, originally being a mimetic weed of wheat.

Vavilovian mimicry describes weeds which come to share characteristics with a domesticated plant through artificial selection. It is named after Russian botanist and geneticist Nikolai Vavilov. Selection against the weed may occur either by manually killing the weed, or separating its seeds from those of the crop. The latter process, known as winnowing, can be done manually or by a machine.

Vavilovian mimicry presents an illustration of unintentional (or rather 'anti-intentional') selection by man. While some cases of artificial selection go in the direction desired, such as selective breeding, this case presents the opposite characteristics. Weeders do not want

to select weeds that look increasingly like the cultivated plant, yet there is no other option. A similar problem in agriculture is pesticide. Vavilovian mimics may eventually be domesticated themselves, and Vavilov called these weeds-cum-crops *secondary crops*.

It can be classified as defensive mimicry in that the weed mimics a protected species. This bears strong similarity to Batesian mimicry in that the weed does not share the properties that give the model its protection, and both the model and the dupe (in this case people) are harmed by its presence. There are some key differences, though; in Batesian mimicry the model and signal receiver are enemies (the predator would eat the protected species if could), whereas here the crop and its human growers are in a mutualistic relationship: the crop benefits from being dispersed and protected by people, despite being eaten by them. In fact, the crop's only 'protection' relevant here is its usefulness to humans. Secondly, the weed is not eaten, but simply destroyed. The only motivation for killing the weed is its effect on crop yields. Finally, this type of mimicry does not occur in ecosystems unaltered by humans.

One case is *Echinochloa oryzoides*, a species of grass which is found as a weed in rice (*Oryza sativa*) fields. The plant looks similar to rice and its seeds are often mixed in rice and difficult to separate. This close similarity was enhanced by the weeding process which is a selective force that increases the similarity of the weed in each subsequent generation.

Protective egg decoys

Unlike the above forms of mimicry, *Gilbertian mimicry* involves only two species. The potential host/prey drives away its parasite/predator by mimicking it, the reverse of host-parasite aggressive mimicry. It was coined by Pasteur as a term for such rare mimicry systems, and is named after the American ecologist Lawrence E. Gilbert.

This form of protective mimicry occurs in the genus *Passiflora*. The leaves of this plant contain toxins which deter herbivorous animals, however some *Heliconius* butterfly larvae have evolved enzymes which break down these toxins, allowing them to specialize on this genus. This has created further selection pressure on the host plants, which have evolved stipules that mimic mature *Heliconius* eggs near the point of hatching. These butterflies tend to avoid laying eggs near each existing ones, which helps avoid exploitative intraspecific competition between caterpillars—those that lay on vacant leaves provide their offspring with a greater chance of survival. Additionally, most *Heliconius* larvae are cannibalistic, meaning those leaves with older eggs will hatch first and eat the new arrivals. Thus, it seems such plants have evolved egg dummies due to these grazing herbivore enemies. The decoy eggs are also nectaries though, attracting predators of the caterpillars such as ants and wasps. The extent of their mimetic function is therefore slightly more difficult to assess.

The use of eggs is not essential to this system, only the species composition and protective function. Many other forms of mimicry also involve eggs, such as cuckoo eggs

mimicking those of their host (the reverse of this situation), or plants seeds (often those with an elaiosome) being dispersed by ants, who treat them as they would their own eggs.

Protective mimicry within a species

Monarch caterpillars, shown feeding, vary in toxicity depending on their diet.

Browerian mimicry, named after Lincoln P. Brower and Jane Van Zandt Brower, is a form of *automimicry*; where the model belongs to the same species as the mimic. This is the analogue of Batesian mimicry within a single species, and occurs when there is a palatability spectrum within a population. One example is Monarch Butterflies (*Danaus plexippus*), which feed on milkweed species of varying toxicity. This species stores toxins from its host plant, which are maintained even in the adult (imago) form. As the levels of toxin will vary depending on diet during the larval stage, some individuals will be more toxic than others. The less palatable organisms will therefore be mimics of the more dangerous individuals, with their likeness already perfected. This need not be the case however; in sexually dimorphic species one sex may be more of a threat than the other, which could mimic the protected sex. Evidence for this possibility is provided by the behavior of a monkey from Gabon, which regularly ate male moths of the genus *Anaphe*, but promptly stopped after it tasted a noxious female.

Aggressive

Aggressive mimicry describes predators (or parasites) which share the same characteristics as a harmless species, allowing them to avoid detection by their prey (or host). The mimic may resemble the prey or host itself, or another organism which is either neutral or beneficial to the signal receiver. In this class of mimicry the model may be affected negatively, positively or not at all. Just as parasites can be treated as a form of predator, host-parasite mimicry is treated here as a subclass of aggressive mimicry.

The mimic may have a particular significance for duped prey. One such case is spiders, amongst which aggressive mimicry is quite common in both luring prey and stealthily approaching predators. One case is the Golden Orb Weaver (*Nephila clavipes*), which spins a conspicuous golden colored web in well-lit areas. Experiments show that bees are able to associate the webs with danger when the yellow pigment is not present, as occurs in less well-lit areas where the web is much harder to see. Other colors were also learned and avoided, but bees seemed least able to effectively associate yellow pigmented webs with danger. Yellow is the color of many nectar bearing flowers, however, so perhaps avoiding yellow is not worth while. Another form of mimicry is based not on color but pattern. Species such as *Argiope argentata* employ prominent patterns in the middle of their webs, such as zigzags. These may reflect ultraviolet light, and mimic the pattern seen in many flowers known as nectar guides. Spiders change their web day to day, which can be explained by bee's ability to remember web patterns. Bees are able to associate a certain pattern with a spatial location, meaning the spider must spin a new pattern regularly or suffer diminishing prey capture.

Another case is where males are lured towards what would seem to be a sexually receptive female; the model in this situation being the same species as the dupe. Beginning in the 1960s, James E. Lloyd's investigation of female fireflies of the genus *Photuris* revealed they emit the same light signals that females of the genus *Photinus* use as a mating signal. Further research showed male fireflies from several different genera are attracted to these "femmes fatales", and are subsequently captured and eaten. Female signals are based on that received from the male, each female having a repertoire of signals matching the delay and duration of the female of the corresponding species. This mimicry may have evolved from non-mating signals that have become modified for predation.

The Spotted Predatory Katydid (*Chlorobalius leucoviridis*) is an acoustic aggressive mimic of cicadas.

The listrosceline katydid *Chlorobalius leucoviridis* of inland Australia is capable of attracting male cicadas of the Tribe Cicadettini by imitating the species-specific reply clicks of sexually receptive female cicadas. This example of acoustic aggressive mimicry is similar to the *Photuris* firefly case in that the predator's mimicry is remarkably versatile – playback experiments show that *C. leucoviridis* is able to attract males of many cicada species, including Cicadettine cicadas from other continents, even though cicada mating signals are species-specific.

Some carnivorous plants may also be able to increase their rate of capture through mimicry.

Two Bluestreak cleaner wrasse cleaning a Potato grouper, *Epinephelus tukula*

Luring is not a necessary condition however, as the predator will still have a significant advantage by simply not being identified as such. They may resemble a mutualistic symbiont or a species of little relevance to the prey.

A case of the former situation is a species of cleaner fish and its mimic, though in this example the model is greatly disadvantaged by the presence of the mimic. Cleaner fish are the allies of many other species, which allow them to eat their parasites and dead skin. Some allow the cleaner to venture inside their body to hunt these parasites. However, one species of cleaner, the Bluestreak cleaner wrasse (*Labroides dimidiatus*), is the unknowing model of a mimetic species, the Sabre-toothed blenny (*Aspidontus taeniatus*). This wrasse, shown to the left cleaning a grouper of the genus *Epinephelus*, resides in coral reefs in the Indian and the Pacific Oceans, and is recognized by other fishes who then allow it to clean them. Its imposter, a species of blenny, lives in the Indian Ocean and not only looks like it in terms of size and coloration, but even mimics the cleaner's 'dance'. Having fooled its prey into letting its guard down, it then bites it, tearing off a piece of its fin before fleeing the scene. Fish grazed upon in this fashion soon learn to distinguish mimic from model, but because the similarity is close between the two they become much more cautious of the model as well, such that both are affected. Due to victim's ability to discriminate between foe and helper, the blennies have evolved close similarity, right down to the regional level.

Another interesting example that does not involve any luring is the Zone-tailed Hawk, which resembles the Turkey Vulture. It flies amongst the vultures, suddenly breaking from the formation and ambushing its prey. Here the hawk's presence is of no evident significance to the vultures, affecting them neither negatively or positively.

Parasites

Parasites can also be aggressive mimics, though the situation is somewhat different from those outlined above.

Some of the predators described have a feature that draws prey, and parasites can also mimic their host's natural prey, but are eaten themselves, a pathway into their host. *Leucochloridium*, a genus of flatworm, matures in the digestive system of songbirds, their eggs then passing out of the bird via the feces. They are then taken up by *Succinea*, a terrestrial snail. The eggs develop in this intermediate host, and then must find of a suitable bird to mature in. As the host birds do not eat snails, so the sporocyst has another strategy to reach its host's intestine. They are brightly colored and move in a pulsating fashion. A sporocyst-sac pulsates in the snail's eye stalks, coming to resemble an irresistible meal for a songbird. In this way, it can bridge the gap between hosts, allowing it to complete its life cycle. A nematode (*Myrmeconema neotropicum*) changes the colour of the abdomen of workers of the canopy ant *Cephalotes atratus* to make it appear like the ripe fruits of *Hyeronima alchorneoides*. It also changes the behaviour of the ant so that the gaster (rear part) is held raised. This presumably increases the chances of the ant being eaten by birds. The droppings of birds are collected by other ants and fed to their brood, thereby helping to spread the nematode.

In an unusual case, planidium larvae of some beetles of the genus *Meloe* will form a group and produce a pheromone that mimics the sex attractant of its host bee species; when the male bee arrives and attempts to mate with the mass of larvae, they climb onto his abdomen, and from there transfer to a female bee, and from there to the bee nest to parasitize the bee larvae.

Host-parasite mimicry is a two species system where a parasite mimics its own host. Cuckoos are a canonical example of brood parasitism, a form of kleptoparasitism where the mother has its offspring raised by another unwitting organism, cutting down the biological mother's parental investment in the process. The ability to lay eggs which mimic the host eggs is the key adaptation. The adaptation to different hosts is inherited through the female line in so-called gentes. Cases of *intraspecific* brood parasitism, where a female lays in conspecific's nest, as illustrated by the Goldeneye duck (*Bucephala clangula*), do not represent a case of mimicry.

Reproductive

Reproductive mimicry occurs when the actions of the dupe directly aid in the mimic's reproduction. This is common in plants, which may have deceptive flowers that do not provide the reward they would seem to. Other forms of mimicry have a reproductive

component, such as Vavilovian mimicry involving seeds, and brood parasitism, which also involves aggressive mimicry.

Mimicry of flowers

Bakerian mimicry, named after Herbert G. Baker, is a form of automimicry where female flowers mimic male flowers of their own species, cheating pollinators out of a reward. This reproductive mimicry may not be readily apparent as members of the same species may still exhibit some degree of sexual dimorphism. It is common in many species of Caricaceae.

Like Bakerian mimicry, *Dodsonian mimicry* is a form of reproductive floral mimicry, but the model belongs to a different species than the mimic. The name refers to Calaway H. Dodson. By providing similar sensory signals as the model flower, it can lure its pollinators. Like Bakerian mimics, no nectar is provided. *Epidendrum ibaguense* of the family Orchidaceae resembles flowers of *Lantana camara* and *Asclepias curassavica*, and is pollinated by Monarch Butterflies and perhaps hummingbirds. Similar cases are seen in some other species of the same family. The mimetic species may still have pollinators of its own though, for example a lamellicorn beetle which usually pollinates correspondingly colored *Cistus* flowers is also known to aid in pollination of *Ophrys* species that are normally pollinated by bees.

Pseudocopulation

The Fly Orchid (*Ophrys insectifera*)

Pseudocopulation occurs when a flower mimics a female of a certain insect species, the males of which try to copulate with it. This is much like the aggressive mimicry in fireflies described above, but with a much more benign outcome for the pollinator. This form of mimicry has been called *Pouyannian mimicry*, after Pouyanne, who first described the phenomenon. It is most common in orchids which mimic females of the order Hymenoptera (generally bees and wasps), and may account for around 60% of pollinations. Depending on the morphology of the flower, a pollen sac called a pollinia is attached to the head or abdomen of the male. This is then transferred to the stigma of the next flower the male tries to inseminate, resulting in pollination. Visual mimicry is the

most obvious sign of this deception for humans, but the visual aspect may be minor or non-existent. It is the senses of touch and olfaction that are most important.

Inter-sexual mimicry

Inter-sexual mimicry occurs when individuals of one sex in a species mimic members of the opposite sex. An example is the three male forms of the marine isopod, *Paracerceis sculpta*. Alpha males are the largest and guard a harem of females. Beta males mimic females and manage to enter the harem of females without being detected by the alpha males allowing them to mate. Gamma males are the smallest males and mimic juveniles. This also allows them to mate with the females without the alpha males detecting them. Some male Australian Giant Cuttlefish also mimic females, allowing them to mate undetected by other males.

Automimicry

Automimicry or *intraspecific mimicry* occurs within a single species, one case being where one part of an organism's body resembles another part. Examples include snakes in which the tail resembles the head and show behavior such as moving backwards to confuse predators and insects and fishes with eyespots on their hind ends to resemble the head. The term is also used when the mimic imitates other morphs within the same species. When males mimic females or *vice versa* this may be referred to as sexual mimicry.

Examples:

- Many insects have filamentous "tails" at the ends of their wings which are combined with patterns of markings on the wings themselves to create a "false head" which misdirects predators (e.g., hairstreak butterflies).
- Several pygmy owls bear "false eyes" on the back of their head to fool predators into believing the owl is alert to their presence.
- The yellow throated males of the Common Side-blotched Lizard use a 'sneaking' strategy in mating. They look and behave like unreceptive females. This strategy is effective against 'usurper' males with orange throats, but ineffective against blue throated 'guarder' males, which will chase them away.
- Female hyenas have pseudo-penises which make them look like males.

Other

Some forms of mimicry do not fit easily within the classification given above.

Owl butterflies (genus *Caligo*) bear eye-spots on the underside of their wings; if turned upside-down, their undersides resemble the face of an owl (such as the Short-eared Owl or the Tropical Screech Owl) for which in turn the butterfly predators – small lizards and birds – would be fooled. Thus it has been supposed that the eye-spots are a form of Batesian mimicry. However, the pose in which the butterfly resembles an owl's head is

not normally adopted in life. Research suggests that eye-spots are not a form of mimicry and do not deter predators because they look like eyes. Rather the conspicuous contrast in the patterns on the wings deter predators.

Another case is floral mimicry induced by the discomycete fungus *Monilinia vaccinii-corymbosi*. In this unusual case, a fungal plant pathogen infects leaves of blueberries, causing them to secrete sugary substances including glucose and fructose, in effect mimicking the nectar of flowers. To the naked eye the leaves do not look like flowers, yet strangely they still attract pollinating insects like bees. As it turns out, the sweet secretions are not the only cues—the leaves also reflect ultraviolet, which is normally absorbed by the plant's leaves. Ultraviolet light is also employed by the host's flowers as a signal to insects, which have visual systems quite capable of picking up this low wavelength (300–400 nm) radiation. The fungus is then transferred to the ovaries of the flower where it produces mummified, inedible berries, which overwinter before infecting new plants. This case is unusual in that the fungus benefits from the deception, but it is the leaves which act as mimics, being harmed in the process. It bears similarity to host-parasite mimicry, but the host does not receive the signal. It also has a little in common with automimicry, but the plant does not benefit from the mimicry, and the action of the pathogen is required to produce it.

Evolution

Ctenomorphodes chronus mimicking a eucalyptus twig

It is widely accepted that mimicry evolves as a positive adaptation; that is, the mimic gains fitness gradually *via* convergent evolution which results in resemblance to another species. The lepidopterist (and sometime author) Vladimir Nabokov argued that although natural selection might stabilize a "mimic" form, it would not be necessary to create it. It may be that much of insect mimicry, including the Viceroy/Monarch mimicry, results from similar self-organizing processes, and thus the tendency for convergence by chance would be high.

The most widely accepted model used to explain the evolution of mimicry in butterflies is the two-step hypothesis. In this model the first step involves mutation in modifier genes that regulate a complex cluster of linked genes associated with large changes in morphology. The second step consists of selections on genes with smaller phenotypic effects and this leading to increasing closeness of resemblance. This model is supported

by empirical evidence that suggests that there are only a few single point mutations that cause large phenotypic effects while there are numerous others that produce smaller effects. Some regulatory elements are now known to be involved in a supergene that is involved in the development of butterfly color patterns. Computational simulations of population genetics have also supported this idea.

Chapter 2

Polymorphism (Biology)

Light-morph Jaguar (typical)

Dark-morph or melanistic Jaguar (about 6% of the South American population)

Polymorphism in biology occurs when two or more clearly different phenotypes exist in the same population of a species — in other words, the occurrence of more than one *form* or *morph*. In order to be classified as such, morphs must occupy the same habitat at the same time and belong to a panmictic population (one with random mating).

Polymorphism is common in nature; it is related to biodiversity, genetic variation and adaptation; it usually functions to retain variety of form in a population living in a varied environment. The most common example is sexual dimorphism, which occurs in many organisms. Other examples are mimetic forms of butterflies, and human haemoglobin and blood types.

Polymorphism results from evolutionary processes, as does any aspect of a species. It is heritable, and is modified by natural selection. In polyphenism, an individual's genetic make-up allows for different morphs, and the switch mechanism that determines which morph is shown is environmental. In *genetic polymorphism* the genetic make-up determines the morph. Ants exhibit both types in a single population.

Polymorphism as described here involves morphs of the phenotype. The term is also used somewhat differently by molecular biologists to describe certain point mutations in the genotype, such as SNPs.

Terminology

Although in general use polymorphism is quite a broad term, in biology it has been given a specific meaning.

- The term omits characters showing *continuous variation* (such as weight), even though this has a heritable component. Polymorphism deals with forms in which the variation is discrete (discontinuous) or strongly bimodal or polymodal.
- Morphs must occupy the same habitat at the same time: this excludes geographical races and seasonal forms. The use of the words *morph* or *polymorphism* for what is a visibly different *geographical race or variant* is common, but incorrect. The significance of geographical variation is in that it may lead to allopatric speciation, whereas true polymorphism takes place in panmictic populations.
- The term was first used to describe *visible forms*, but nowadays it has been extended to include *cryptic morphs*, for instance blood types, which can be revealed by a test.
- Rare variations are not classified as polymorphisms; and mutations by themselves do not constitute polymorphisms. To qualify as a polymorphism there has to be some kind of balance between morphs underpinned by inheritance. The criterion is that the frequency of the *least* common morph is too high simply to be the result of new mutations or, as a rough guide, that it is greater than 1 percent (though that is far higher than any normal mutation rate for a single allele).

Nomenclature

Polymorphism crosses several discipline boundaries, including ecology and genetics, evolution theory, taxonomy, cytology and biochemistry. Different disciplines may give the same concept different names, and different concepts may be given the same name. For example, there are the terms established in ecological genetics by E.B. Ford (1975), and for classical genetics by John Maynard Smith (1998). The shorter term *morphism* may be more accurate than *polymorphism*, but is not often used. It was the preferred term of the evolutionary biologist Julian Huxley (1955).

Various synonymous terms exist for the various polymorphic forms of an organism. The most common are *morph* and *morpha*, while a more formal term is *morphotype*. *Form* and *phase* are sometimes also used, but are easily confused in zoology with, respectively, "form" in a population of animals, and "phase" as a color or other change in an organism due to environmental conditions (temperature, humidity, etc). Phenotypic traits and characteristics are also possible descriptions, though that would imply just a limited aspect of the body.

In the taxonomic nomenclature of zoology, the word "morpha" plus a Latin name for the morph can be added to a binomial or trinomial name. However, this invites confusion with geographically-variant ring species or subspecies, especially if polytypic. Morphs have no formal standing in the ICZN. In botanical taxonomy, the concept of morphs is

represented with the terms "variety", "subvariety" and "form", which are formally regulated by the ICBN. Horticulturalists sometimes confuse this usage of "variety" both with cultivar ("variety" in viticultural usage, rice agriculture jargon, and informal gardening lingo) and with the legal concept "plant variety" (protection of a cultivar as a form of intellectual property).

Ecology

Selection, whether natural or artificial, changes the frequency of morphs within a population; this occurs when morphs reproduce with different degrees of success. A genetic (or *balanced*) polymorphism usually persists over many generations, maintained by two or more opposed and powerful selection pressures. Diver (1929) found banding morphs in *Cepaea nemoralis* could be seen in pre-fossil shells going back to the Mesolithic Holocene. Apes have similar blood groups to humans: human and chimpanzee blood, with compatible blood groups, can be exchanged through transfusion (Great Ape Project). This suggests rather strongly that this kind of polymorphism is quite ancient, at least as far back as the last common ancestor of the apes and man, and possibly even further.

The white morph of the monarch in Hawaii is partly a result of apostatic selection

The relative proportions of the morphs may vary; the actual values are determined by the effective fitness of the morphs at a particular time and place. The mechanism of heterozygote advantage assures the population of some alternative alleles at the locus or loci involved. Only if competing selection disappears will an allele disappear. However, heterozygote advantage is not the only way a polymorphism can be maintained. Apostatic selection, whereby a predator consumes a common morph whilst overlooking rarer

morphs is possible and does occur. This would tend to preserve rarer morphs from extinction.

A polymorphic population does not initiate speciation; nor does it prevent speciation. It has little or nothing to do with species splitting. However, *it has a lot to do with the adaptation of a species to its environment*, which may vary in colour, food supply, predation and in many other ways. Polymorphism is one good way the opportunities get to be used; it has survival value, and the selection of modifier genes may reinforce the polymorphism.

Polymorphism and niche diversity

G. Evelyn Hutchinson, a founder of niche research, commented "It is very likely from an ecological point of view that all species, or at least all common species, consist of populations adapted to more than one niche". He gave as examples sexual size dimorphism and mimicry. In many cases where the male is short-lived and smaller than the female, he does not compete with her during her late pre-adult and adult life. Size difference may permit both sexes to exploit different niches. In elaborate cases of mimicry, such as the African butterfly *Papilio dardanus*, female morphs mimic a range of distasteful models, often in the same region. The fitness of each type of mimic decreases as it becomes more common, so the polymorphism is maintained by frequency-dependent selection. Thus the efficiency of the mimicry is maintained in a much increased total population.

The switch

The mechanism which decides which of several morphs an individual displays is called the *switch*. This switch may be genetic, or it may be environmental. Taking sex determination as the example, in man the determination is genetic, by the XY sex-determination system. In Hymenoptera (ants, bees and wasps), sex determination is by haplo-diploidy: the females are all diploid, the males are haploid. However, in some animals an environmental trigger determines the sex: alligators are a famous case in point. In ants the distinction between workers and guards is environmental, by the feeding of the grubs. Polymorphism with an environmental trigger is called polyphenism.

The polyphenic system does have a degree of environmental flexibility not present in the genetic polymorphism. However, such environmental triggers are the less common of the two methods.

Investigative methods

Investigation of polymorphism requires a coming together of field and laboratory technique. In the field:

- detailed survey of occurrence, habits and predation
- selection of an ecological area or areas, with well-defined boundaries

- capture, mark, release, recapture data
- relative numbers and distribution of morphs
- estimation of population sizes

And in the laboratory:

- genetic data from crosses
- population cages
- chromosome cytology if possible
- use of chromatography or similar techniques if morphs are cryptic (for example, biochemical)

Both types of work are equally important. Without proper field-work the significance of the polymorphism to the species is uncertain; without laboratory breeding the genetic basis is obscure. Even with insects the work may take many years; examples of Batesian mimicry noted in the nineteenth century are still being researched.

Genetics

Genetic polymorphism

Since all polymorphism has a genetic basis, *genetic polymorphism* has a particular meaning:

- Genetic polymorphism is the simultaneous occurrence in the same locality of two or more discontinuous forms in such proportions that the rarest of them cannot be maintained just by recurrent mutation.

The definition has three parts: a) sympatry: one interbreeding population; b) discrete forms; and c) not maintained just by mutation.

Genetic polymorphism is actively and steadily maintained in populations by natural selection, in contrast to *transient polymorphisms* where a form is progressively replaced by another. By definition, genetic polymorphism relates to a balance or equilibrium between morphs. The mechanisms that conserve it are types of balancing selection.

Mechanisms of balancing selection

- Heterosis (or heterozygote advantage): "Heterosis: the heterozygote at a locus is fitter than either homozygote".

- Frequency dependent selection: The fitness of a particular phenotype is dependent on its frequency relative to other phenotypes in a given population. Example: prey switching, where rare morphs of prey are actually fitter due to predators concentrating on the more frequent morphs.

- Fitness varies in time and space. Fitness of a genotype may vary greatly between larval and adult stages, or between parts of a habitat range.

- Selection acts differently at different levels. The fitness of a genotype may depend on the fitness of other genotypes in the population: this covers many natural situations where the best thing to do (from the point of view of survival and reproduction) depends on what other members of the population are doing at the time.

Pleiotropism

Most genes have more than one effect on the phenotype of an organism (pleiotropism). Some of these effects may be visible, and others cryptic, so it is often important to look beyond the most obvious effects of a gene to identify other effects. Cases occur where a gene affects an unimportant visible character, yet a change in fitness is recorded. In such cases the gene's other (cryptic or 'physiological') effects may be responsible for the change in fitness.

"If a neutral trait is pleiotropically linked to an advantageous one, it may emerge because of a process of natural selection. It was selected but this doesn't mean it is an adaptation. The reason is that, although it was selected, there was no selection for that trait."

Epistasis

Epistasis occurs when the expression of one gene is modified by another gene. For example, gene A only shows its effect when allele B1 (at another locus) is present, but not if it is absent. This is one of the ways in which two or more genes may combine to produce a coordinated change in more than one characteristic (for instance, in mimicry). Unlike the supergene, epistatic genes do not need to be closely linked or even on the same chromosome.

Both pleiotropism and epistasis show that a gene need not relate to a character in the simple manner that was once supposed.

The origin of supergenes

Although a polymorphism can be controlled by alleles at a single locus (e.g. human ABO blood groups), the more complex forms are controlled by supergenes consisting of several tightly linked genes on a single chromosome. Batesian mimicry in butterflies and heterostyly in angiosperms are good examples. There is a long-standing debate as to how this situation could have arisen, and the question is not yet resolved.

Whereas a gene family (several tighly linked genes performing similar or identical functions) arises by duplication of a single original gene, this is usually not the case with supergenes. In a supergene some of the constituent genes have quite distinct functions, so they must have come together under selection. This process might involve suppression of

crossing-over, translocation of chromosome fragments and possibly occasional cistron duplication. That crossing-over can be suppressed by selection has been known for many years.

Debate has centred round the question of whether the component genes in a super-gene could have started off on separate chromosomes, with subsequent reorganization, or if it is necessary for them to start on the same chromosome. Originally, it was held that chromosome rearrangement would play an important role. This explanation was accepted by E. B. Ford and incorporated into his accounts of ecological genetics.

However, today many believe it more likely that the genes start on the same chromosome. They argue that supergenes arose *in situ*. This is known as Turner's sieve hypothesis. John Maynard Smith agreed with this view in his authoritative textbook, but the question is still not definitively settled.

Relevance for evolutionary theory

Polymorphism was crucial to research in ecological genetics by E. B. Ford and his co-workers from the mid-1920s to the 1970s (similar work continues today, especially on mimicry). The results had a considerable effect on the mid-century evolutionary synthesis, and on present evolutionary theory. The work started at a time when natural selection was largely discounted as the leading mechanism for evolution, continued through the middle period when Sewall Wright's ideas on drift were prominent, to the last quarter of the 20th century when ideas such as Kimura's neutral theory of molecular evolution was given much attention. The significance of the work on ecological genetics is that it has shown how important selection is in the evolution of natural populations, and that selection is a much stronger force than was envisaged even by those population geneticists who believed in its importance, such as Haldane and Fisher.

In just a couple of decades the work of Fisher, Ford, Arthur Cain, Philip Sheppard and Cyril Clarke promoted natural selection as the primary explanation of variation in natural populations, instead of genetic drift. Evidence can be seen in Mayr's famous book *Animal Species and Evolution*, and Ford's *Ecological Genetics*. Similar shifts in emphasis can be seen in most of the other participants in the evolutionary synthesis, such as Stebbins and Dobzhansky, though the latter was slow to change.

Kimura drew a distinction between molecular evolution, which he saw as dominated by selectively neutral mutations, and phenotypic characters, probably dominated by natural selection rather than drift. This does not conflict with the account of polymorphism given here, though most of the ecological geneticists believed that evidence would gradually accumulate against his theory.

Examples

Sexual dimorphism

We meet genetic polymorphism daily, since our species (like most other eukaryotes) uses sexual reproduction, and of course, the sexes are differentiated. However, even if the sexes were identical in superficial appearance, the division into two sexes is a dimorphism, albeit cryptic. This is because the phenotype of an organism includes its sexual organs and its chromosomes, and all the behaviour associated with reproduction. So research into sexual dimorphism has addressed two issues: first, the advantage of sex in evolutionary terms; second, the role of visible sexual differentiation.

The system is relatively stable (with about half of the population of each sex) and heritable, usually by means of sex chromosomes. Every aspect of this everyday phenomenon bristles with questions for the theoretical biologist. Why is the ratio ~50/50? How could the evolution of sex occur from an original situation of asexual reproduction, which has the advantage that every member of a species could reproduce? Why the visible differences between the sexes? These questions have engaged the attentions of biologists such as Charles Darwin, August Weismann, Ronald Fisher, George C. Williams, John Maynard Smith and W. D. Hamilton, with varied success.

Of the many issues involved, there is widespread agreement on the following: the advantage of sexual and hermaphroditic reproduction over asexual reproduction lies in the way recombination increases the genetic diversity of the ensuing population.[p234ch7] The advantage of sexual reproduction over hermaphroditic is not so clear. In forms that have two separate sexes, same sex combinations are excluded from mating which decreases the amount of diversity compared with hermaphrodites by at least twice. So, why are almost all progressive species bi-sexual, considering the asexual process is more efficient and simple, whilst hermaphrodites produce a more diversified progeny? It has been suggested that differentiation into two sexes has evolutionary advantages allowing changes to concentrate in the male part of the population and at the same time preserving the existing genotype distribution in the females. This enables the population to better meet the challenges of infection, parasitism, predation and other hazards of the varied environment.

Human polymorphisms

Apart from sexual dimorphism, there are many other examples of human genetic polymorphisms. Infectious disease has been a major factor in human mortality, and so has affected the evolution of human populations. Evidence is now strong that many polymorphisms are maintained in human populations by balancing selection.

Human blood groups

All the common blood types, such as the ABO system, are genetic polymorphisms. Here we see a system where there are more than two morphs: the phenotypes are A, B, AB and

O are present in all human populations, but vary in proportion in different parts of the world. The phenotypes are controlled by multiple alleles at one locus. These polymorphisms are seemingly never eliminated by natural selection; the reason came from a study of disease statistics.

Statistical research has shown that the various phenotypes are more, or less, likely to suffer a variety of diseases. For example, an individual's susceptibility to cholera (and other diarrheal infections) is correlated with their blood type: those with type O blood are the most susceptible, while those with type AB are the most resistant. Between these two extremes are the A and B blood types, with type A being more resistant than type B. This suggests that the pleiotropic effects of the genes set up opposing selective forces, thus maintaining a balance. Geographical distribution of blood groups (the differences in gene frequency between populations) is broadly consistent with the classification of "races" developed by early anthropologists on the basis of visible features.

Sickle-cell anaemia

Such a balance is seen more simply in sickle-cell anaemia, which is found mostly in tropical populations in Africa and India. An individual homozygous for the recessive sickle haemoglobin, HgbS, has a short expectancy of life, whereas the life expectancy of the standard haemoglobin (HgbA) homozygote and also the heterozygote is normal (though heterozygote individuals will suffer periodic problems). The sickle-cell variant survives in the population because the heterozygote is resistant to malaria and the malarial parasite kills a huge number of people each year. This is balancing selection or genetic polymorphism, balanced between fierce selection against homozygous sickle-cell sufferers, and selection against the standard HgbA homozygotes by malaria. The heterozygote has a permanent advantage (a higher fitness) so long as malaria exists; and it has existed as a human parasite for a long time. Because the heterozygote survives, so does the HgbS allele survive at a rate much higher than the mutation rate.

Duffy system

The Duffy antigen is a protein located on the surface of red blood cells, encoded by the *FY (DARC)* gene. The protein encoded by this gene is a non-specific receptor for several chemokines, and is the known entry-point for the human malarial parasites *Plasmodium vivax* and *Plasmodium knowlesi*. Polymorphisms in this gene are the basis of the Duffy blood group system.

In humans, a mutant variant at a single site in the FY cis-regulatory region abolishes all expression of the gene in erythrocyte precursors. As a result, homozygous mutants are strongly protected from infection by *P. vivax*, and a lower level of protection is conferred on heterozygotes. The variant has apparently arisen twice in geographically distinct human populations, in Africa and Papua New Guinea. It has been driven to high frequencies on at least two haplotypic backgrounds within Africa. Recent work indicates a similar, but not identical, pattern exists in baboons (*Papio cynocephalus*), which suffer

a mosquito-carried malaria-like pathogen, *Hepatocystis kochi*. Researchers interpret this as a case of convergent evolution.

G6PD

G6PD (Glucose-6-phosphate dehydrogenase) human polymorphism is also implicated in malarial resistance. G6PD alleles with reduced activity are maintained at a high level in endemic malarial regions, despite reduced general viability. Variant A (with 85% activity) reaches 40% in sub-Saharan Africa, but is generally <1% outside Africa and the Middle East.

Cystic fibrosis

Cystic fibrosis, a congenital defect which affects about one in 2000 children, is caused by a mutant form of the CF transmembrane regulator gene, CFTR. The transmission is Mendelian: the normal gene is dominant, so all heterozygotes are healthy, but those who inherit two mutated genes have the condition. The mutated allele is present in about 1:25 of the population (mostly heterozygotes), which is much higher than expected from the rate of mutation alone. Sufferers from this disease have shortened life expectancy (and males are usually sterile if they survive), and the disease was effectively lethal in pre-modern societies. The incidence of the disease varies greatly between ethnic groups, but is highest in Caucasian populations.

Although over 1500 mutations are known in the CFTR gene, by far the most common mutant is DF508. This mutant is being kept at a high level in the population despite the lethal or near-lethal effects of the mutant homozygote. It seems that some kind of heterozygote advantage is operating. Early theories that the heterozygotes might enjoy increased fertility have not been borne out. Present indications are that the bacterium which causes typhoid fever enters cells using CFTR, and experiments with mice suggest that heterozygotes are resistant to the disease. If the same were true in humans, then heterozygotes would have had a great advantage during typhoid epidemics. Cystic fibrosis is a prime target for gene therapy research.

Human taste morphisms

A famous puzzle in human genetics is the genetic ability to taste phenylthiourea (phenylthiocarbamide or PTC), a morphism which was discovered in 1931. This substance, which to some of us is bitter, and to others tasteless, is of no great significance in itself, yet it is a genetic dimorphism. Because of its high frequency (which varies in different ethnic groups) it must be connected to some function of selective value. The ability to taste PTC itself is correlated with the ability to taste other bitter substances, many of which are toxic. Indeed, PTC itself is toxic, though not at the level of tasting it on litmus paper. Variation in PTC perception may reflect variation in dietary preferences throughout human evolution, and might correlate with susceptibility to diet-related diseases in modern populations. There is a statistical correlation between PTC tasting and liability to thyroid disease.

Fisher, Ford and Huxley tested orangutans and chimpanzees for PTC perception with positive results, thus demonstrating the long-standing existence of this dimorphism. The recently identified PTC gene, which accounts for 85% of the tasting variance, has now been analysed for sequence variation with results which suggest selection is maintaining the morphism.

Lactose tolerance/intolerance

The ability to metabolize lactose, a sugar found in milk and other dairy products, is a prominent dimorphism that has been linked to recent human evolution.

MHC molecules

The genes of the major histocompatibility complex (MHC) are highly polymorphic, and this diversity plays a very important role in resistance to pathogens. This is true for other species as well.

The cuckoo

Reed warbler feeding a cuckoo chick (*Cuculus canorus*)

Over fifty species in this family of birds practise brood parasitism; the details are best seen in the British or European cuckoo (*Cuculus canorus*). The female lays 15–20 eggs in a season, but only one in each nest of another bird. She removes some or all of the host's clutch of eggs, and lays an egg which closely matches the host eggs. Although, in Britain, the hosts are always smaller than the cuckoo itself, the eggs she lays are small, and coloured to match the host clutch but thick-shelled. This latter is a defence which protects the egg if the host detects the fraud.

The intruded egg develops exceptionally quickly; when the newly-hatched cuckoo is only ten hours old, and still blind, it exhibits an urge to eject the other eggs or nestlings. It rolls

them into a special depression on its back and heaves them out of the nest. The cuckoo nestling is apparently able to pressure the host adults for feeding by mimicking the cries of the host nestlings. The diversity of the cuckoo's eggs is extraordinary, the forms resembling those of its most usual hosts. In Britain these are:

- Meadow pipit (*Anthus pratensis*): brown eggs speckled with darker brown.
- European robin (*Erithacus rubecula*): whitish-grey eggs speckled with bright red.
- Reed warbler (*Acrocephalus scirpensis*): light dull green eggs blotched with olive.
- Redstart (*Phoenicurus phoenicurus*): clear blue eggs.
- Hedge sparrow (*Prunella modularis*): clear blue eggs, unmarked, not mimicked. This bird is an uncritical fosterer; it tolerates in its nest eggs that do not resemble its own.

Each female cuckoo lays one type only; the same type laid by her mother. In this way female cuckoos are divided into groups (known as *gentes*, singular *gens*), each parasitises the host to which it is adapted. The male cuckoo has its own territory, and mates with females from any gens; thus the population (all gentes) is interbreeding.

The standard explanation of how the inheritance of gens works is as follows. The egg colour is inherited by sex chromosome. In birds sex determination is ZZ/ZW, and unlike mammals, the heterogametic sex is the female. The determining gene (or super-gene) for the inheritance of egg colour is believed to be carried on the W chromosome, which is directly transmitted in the female line. The female behaviour in choosing the host species is set by imprinting after birth, a common mechanism in bird behaviour.

Ecologically, the system of multiple hosts protects host species from a critical reduction in numbers, and maximises the egg-laying capacity of the population of cuckoos. It also extends the range of habitats where the cuckoo eggs may be raised successfully. Detailed work on the Cuckoo started with E. Chance in 1922, and continues to the present day; in particular, the inheritance of gens is still a live issue.

Grove snail

The grove snail, *Cepaea nemoralis*, is famous for the rich polymorphism of its shell. The system is controlled by a series of multiple alleles. The shell colour series is brown (genetically the top dominant trait), dark pink, light pink, very pale pink, dark yellow and light yellow (the bottom or universal recessive trait). Bands may be present or absent; and if present from one to five in number. Unbanded is the top dominant trait, and the forms of banding are controlled by modifier genes.

Grove snail, dark yellow shell with single band.

In England the snail is regularly predated by the song thrush *Turdus philomelos*, which breaks them open on *thrush anvils* (large stones). Here fragments accumulate, permitting researchers to analyse the snails taken. The thrushes hunt by sight, and capture selectively those forms which match the habitat *least well*. Snail colonies are found in woodland, hedgerows and grassland, and the predation determines the proportion of phenotypes (morphs) found in each colony.

Two active grove snails

A second kind of selection also operates on the snail, whereby certain heterozygotes have a physiological advantage over the homozygotes. In addition, apostatic selection is likely, with the birds preferentially taking the most common morph. This is the 'search pattern' effect, where a predominantly visual predator persists in targeting the morph which gave a good result, even though other morphs are available.

Despite the predation, the polymorphism survives in almost all habitats, though the proportions of morphs varies considerably. The alleles controlling the polymorphism form a super-gene with linkage so close as to be nearly absolute. This control saves the population from a high proportion of undesirable recombinants, and it is hypothesised that selection has brought the loci concerned together.

To sum up, in this species predation by birds appears to be the main (but not the only) selective force driving the polymorphism. The snails live on heterogeneous backgrounds, and thrush are adept at detecting poor matches. The inheritance of physiological and cryptic diversity is preserved also by heterozygous advantage in the super-gene. Recent work has included the effect of shell colour on thermoregulation, and a wider selection of possible genetic influences is considered by Cook.

A similar system of genetic polymorphism occurs in the White-lipped Snail *Cepaea hortensis*, a close relative of the grove snail. In Iceland, where there are no song thrushes, a correlation has been established between temperature and colour forms. Banded and brown morphs reach higher temperatures than unbanded and yellow snails. This may be the basis of the physiological selection found in both species of snail.

Scarlet tiger moth

The scarlet tiger moth *Callimorpha* (*Panaxia*) *dominula* (family Arctiidae) occurs in continental Europe, western Asia and southern England. It is a day-flying moth, noxious-tasting, with brilliant warning colour in flight, but cryptic at rest. The moth is colonial in habit, and prefers marshy ground or hedgerows. The preferred food of the larvae is the herb Comfrey (*Symphytum officinale*). In England it has one generation per year.

Callimorpha dominula morpha *typica* with spread wings. The red with black rear wings, revealed in flight, warn of its noxious taste. The front wings are cryptic, covering the rear wings at rest. Here the moth is resting but alert, and has jinked the front wings forward to reveal the warning flash.

The moth is known to be polymorphic in its colony at Cothill, about five miles (8 km) from Oxford, with three forms: the typical homozygote; the rare homozygote (*bimacula*)

and the heterozygote (*medionigra*). It was studied there by Ford and later by Sheppard and their co-workers over many years. Data is available from 1939 to the present day, got by the usual field method of capture-mark-release-recapture and by genetic analysis from breeding in captivity. The records cover gene frequency and population-size for much of the twentieth century.

In this instance the genetics appears to be simple: two alleles at a single locus, producing the three phenotypes. Total captures over 26 years 1939-64 came to 15,784 homozygous *dominula* (i.e. *typica*), 1,221 heterozygous *medionigra* and 28 homozygous *bimacula*. Now, assuming equal viability of the genotypes 1,209 heterozygotes would be expected, so the field results do *not* suggest any heterozygous advantage. It was Sheppard who found that the polymorphism is maintained by selective mating: each genotype preferentially mates with other morphs. This is sufficient to maintain the system despite the fact that in this case the heterozygote has slightly lower viability.

Peppered moth

The peppered moth, *Biston betularia*, is justly famous as an example of a population responding in a heritable way to a significant change in their ecological circumstances. E.B. Ford described it as "one of the most striking, though not the most profound, evolutionary changes ever actually witnessed in nature".

Although the moths are cryptically camouflaged and rest during the day in unexposed positions on trees, they are predated by birds hunting by sight. The original camouflage (or crypsis) seems near-perfect against a background of lichen growing on trees. The sudden growth of industrial pollution in the nineteenth century changed the effectiveness of the moths' camouflage: the trees became blackened by soot, and the lichen died off. In 1848 a dark version of this moth was found in the Manchester area. By 1895 98% of the Peppered Moths in this area were black. This was a rapid change for a species that has only one generation a year.

Biston betularia morpha *typica*, the standard light-coloured Peppered Moth.

Biston betularia morpha *carbonaria*, the melanic Peppered Moth.

In Europe, there are three morphs: the typical white morph (*betularia* or *typica*), and *carbonaria*, the melanic black morph. They are controlled by alleles at one locus, with the carbonaria being dominant. There is also an intermediate or semi-melanic morph *insularia*, controlled by other alleles.

A key fact, not realised initially, is the advantage of the heterozygotes, which survive better than either of the homozygotes. This affects the caterpillars as well as the moths, in spite of the caterpillars being monomorphic in appearance (they are twig mimics). In practice heterozygote advantage puts a limit to the effect of selection, since neither homozygote can reach 100% of the population. For this reason, it is likely that the carbonaria allele was in the population originally, pre-industrialisation, at a low level. With the recent reduction in pollution, the balance between the forms has already shifted back significantly.

Another interesting feature is that the carbonaria had noticeably darkened after about a century. This was seen quite clearly when specimens collected about 1880 were compared with specimens collected more recently: clearly the dark morph has been adjusted by the strong selection acting on the gene complex. This might happen if a more extreme allele was available at the same locus; or genes at other loci might act as modifiers. We do not, of course, know anything about the genetics of the original melanics from the nineteenth century.

This type of industrial melanism has only affected such moths as obtain protection from insect-eating birds by resting on trees where they are concealed by an accurate resemblance to their background (over 100 species of moth in Britain with melanic forms were known by 1980). No species which hide during the day, for instance, among dead leaves, is affected, nor has the melanic change been observed among butterflies.

This is, as advertised in many textbooks, "evolution in action". Much of the work was done by Bernard Kettlewell, whose methods came under scrutiny later on. The entomologist Michael Majerus discussed criticisms made of Kettlewell's experimental methods in his 1998 book *Melanism: Evolution in Action*. This book was misrepresented in some reviews, and the story picked up by creationist campaigners. Judith Hooper, in her controversial book *Of Moths and Men* (2002), implied that Kettlewell's work was fraudulent or incompetent. Careful studies of Kettlewell's surviving papers by Rudge (2005) and Young (2004) found that Hooper's accusation of fraud was unjustified, and that "Hooper does not provide one shred of evidence to support this serious allegation". Majerus himself described *Of Moths and Men* as "littered with errors, misrepresentations, misinterpretations and falsehoods". A suitably restrained summary of latest opinion mostly favours predation as the main selective force.

Current interpretation of the available evidence is that the peppered moth is in fact a valid example of natural selection and adaptation. It illustrates a polymorphic species maintaining adaptation to a varied and sometimes changing environment.

Two-spotted ladybird beetle

red morph

black morph

Adalia bipunctata, the two-spotted ladybird, is highly polymorphic. Its basic form is red with two black spots, but it has many other forms, the most important being melanic, with black elytra and red spots. The curious fact about this morphism is that, although the melanic forms are more common in industrial areas, its maintenance has nothing to do with cryptic camouflage and predation. The Coccinellidae as a whole are highly noxious, and experiments with birds and other predators have found this species quite exceptionally distasteful. Therefore, their colour is warning (aposematic) colouration, and all the morphs are quite conspicuous against green vegetation. The field studies identify differing proportions of morphs at different times of year and in different places, which indicates a high level of selection. However, the basis of that selection is still not known for sure, though many theories have been proposed. Since all the morphs are aposematically coloured, it seems unlikely that the difference between the colour of

morphs is directly under selection. Perhaps pleiotropic effects of the genes acting on colour also affect the beetle's physiology, and hence its relative fitness. A similar polymorphic system is found in many other species in this family: *Harmonia axyridis* is a good example.

Ants

Ants exhibit a range of polymorphisms. First, there is their characteristic haplodiploid sex determination system, whereby all males are haploid, and all females diploid. Second, there is differentiation between both the females and males based mostly on feeding of larvae, which determines, for example, whether the imago is capable of reproduction. Lastly, there is differentiation of size and 'duties' (particularly of females), which are usually controlled by feeding and/or age, but which may sometimes be genetically controlled. Thus the order exhibits both genetic polymorphism and extensive polyphenism.

Hoverfly polymorphism

Xanthogramma pedissiquum, a wasp mimic

Volucella zonaria, a large bumblebee mimic

Mallota sp., a bumblebee mimic

Hoverfly mimics can be seen in almost any garden in the temperate zone. The Syrphidae are a large (5600+ species) family of flies; their imagos feed on nectar and pollen, and are well-known for their mimicry of social hymenoptera. The mimicry is Batesian in nature: hoverflies are palatable but hymenoptera are generally unpalatable and may also be protected by stings and/or armour.

Many social wasp (Vespidae) species exhibit Mullerian mimicry, where a group of unpalatable species benefit from sharing the same kind of warning (aposematic) colouration. Wasps are decidedly noxious: nasty-tasting and with a painful sting. They form a Mullerian 'ring' of similarly coloured models; the wasps are often accompanied by clusters of hover-fly mimics, who tend to arrive at the flowers at a similar time of day, and whose flight pattern is passably similar to wasp flight.

Observers in a garden can see for themselves that hoverfly mimics are quite common, usually many times more common than the models, and are (to our sight) relatively poor mimics, often easy to distinguish from real wasps. However, it has been established in other cases that imperfect mimicry can confer significant advantage to the mimic, especially if the model is really noxious. Also, not only is polymorphism absent from these mimics, it is absent in the wasps also: these facts are presumably connected.

The situation with bumblebees (*Bombus*) is rather different. They too are unpalatable, in the sense of being difficult to eat: their body is covered with setae (like carpet pile) and is

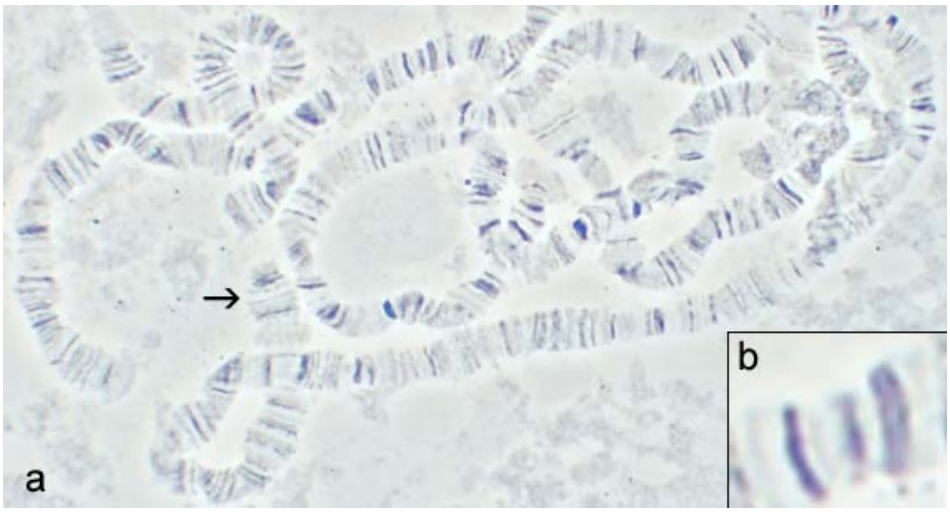
armoured; they are sometimes described as being 'non-food'. Mostler in 1935 carried out tests of their palatability: with the exception of specialist bee-eaters, adults of 19 species of birds ate only 2% of 646 bumblebees presented to them. After various trials, Mostler attributed their avoidance mainly to mechanical difficulties in handling: one young bird took 18 minutes to subdue, kill and eat a bumblebee.

Bumblebees form Mullerian rings of species, and they do often exhibit polymorphism. The hoverfly species mimicking bumblebees are generally accurate mimics, and many of their species are polymorphic. Many of the polymorphisms are different between the sexes, for example by the mimicry being limited to one sex only.

The question is, how can the differences between social wasp mimics and bumblebee mimics be explained? Evidently if model species are common, and have overlapping distributions, they are less likely to be polymorphic. Their mimics are widespread and develop a kind of rough and ready jack-of-all-trades mimicry. But if model species are less common and have patchy distribution they develop polymorphism; and their mimics match them more exactly and are polymorphic also. The issues are currently being investigated.

Chromosome polymorphism in *Drosophila*

In the 1930s Dobzhansky and his co-workers collected *Drosophila pseudoobscura* and *D. persimilis* from wild populations in California and neighbouring states. Using Painter's technique they studied the polytene chromosomes and discovered that the wild populations were polymorphic for chromosomal inversions. All the flies look alike whatever inversions they carry: this is an example of a cryptic polymorphism. Accordingly, Dobzhansky favoured the idea that the morphs became fixed in the population by means of Sewall Wright's drift. However, evidence rapidly accumulated to show that natural selection was responsible:



Drosophila polytene chromosome

1. Values for heterozygote inversions of the third chromosome were often much higher than they should be under the null assumption: if no advantage for any form the number of heterozygotes should conform to N_s (number in sample) = $p^2+2pq+q^2$ where 2pq is the number of heterozygotes.

2. Using a method invented by l'Heretier and Teissier, Dobzhansky bred populations in *population cages*, which enabled feeding, breeding and sampling whilst preventing escape. This had the benefit of eliminating migration as a possible explanation of the results. Stocks containing inversions at a known initial frequency can be maintained in controlled conditions. It was found that the various chromosome types do not fluctuate at random, as they would if selectively neutral, but adjust to certain frequencies at which they become stabilised. With *D. persimilis* he found that the caged population followed the values expected on the Hardy-Weinberg equilibrium when conditions were optimal (which disproved any idea of non-random mating), but with a restricted food supply heterozygotes had a distinct advantage.

3. Different proportions of chromosome morphs were found in different areas. There is, for example, a polymorph-ratio cline in *D. robusta* along an 18-mile (29 km) transect near Gatlinburg, TN passing from 1,000 feet (300 m) to 4,000 feet. Also, the same areas sampled at different times of year yielded significant differences in the proportions of forms. This indicates a regular cycle of changes which adjust the population to the seasonal conditions. For these results selection is by far the most likely explanation.

4. Lastly, morphs cannot be maintained at the high levels found simply by mutation, nor is drift a possible explanation when population numbers are high.

By the time Dobzhansky published the third edition of his book in 1951, he was persuaded that the chromosome morphs were being maintained in the population by the selective advantage of the heterozygotes, as with most polymorphisms. Later he made yet another interesting discovery. One of the inversions, known as PP, was quite rare up to 1946, but by 1958 its proportion had risen to 8%. Not only that, but the proportion was similar over an area of some 200,000 square miles (520,000 km^2) in California. This cannot have happened by migration of PP morphs from, say, Mexico (where the inversion is common) because the rate of dispersal (at less than 2 km/year) is of the wrong order. The change therefore reflected a change in prevailing selection whose basis was not yet known.

Chromosomal polymorphism in general

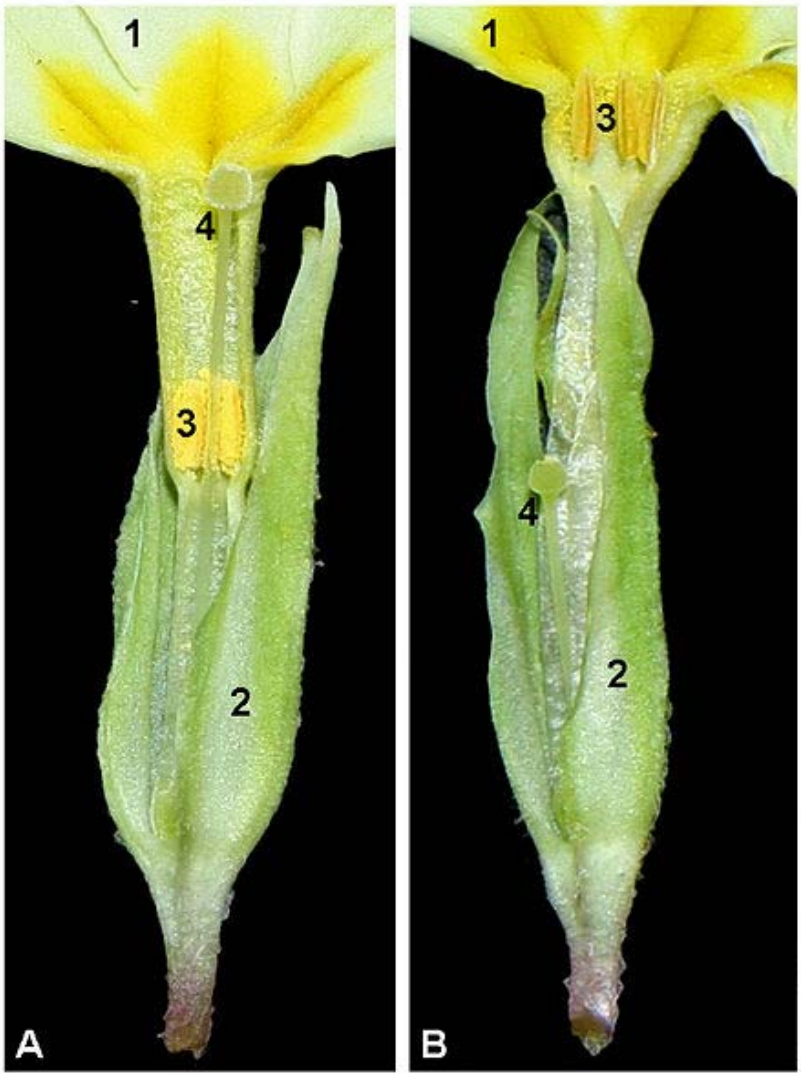
In 1973, M. J. D. White, then at the end of a long career investigating karyotypes, gave an interesting summary of the distribution of chromosome polymorphism.

> "It is extremely difficult to get an adequate idea as to what fraction of the species of eukaryote organisms actually are polymorphic for structural rearrangements of the chromosomes. In Dipterous flies with polytene chromosomes... the figure is somewhere between 60 and 80 percent... In grasshoppers pericentric inversion

polymorphism is shown by only a small number of species. But in this group polymorphism for super-numerary chromosomes and chromosome regions is very strongly developed in many species."
"It is clear that the nature of natural populations is a very complicated subject, and it now appears probable that adaptation of the various genotypes to different ecological niches and frequency-dependent selection are at least as important, and probably more important in many cases, than simple heterosis (in the sense of increased viability or fecundity of the heterozygote)".

This suggests, once again, that polymorphism is a common and important aspect of adaptive evolution in natural populations.

Heterostyly



Dissection of thrum and pin flowers of *Primula vulgaris*

An example of a botanical genetic polymorphism is heterostyly, in which flowers occur in different forms with different arrangements of the pistils and the stamens. The system is called heteromorphic self-incompatibility, and the general 'strategy' of stamens separated from pistils is known as herkogamy.

Pin and thrum heterostyly occurs in dimorphic species of *Primula*, such as *P. vulgaris*. There are two types of flower. The *pin* flower has a long style bearing the stigma at the mouth and the stamens half-way down; and the *thrum* flower has a short style, so the stigma is half-way up the tube and the stamens are at the mouth. So when an insect in search of nectar inserts its proboscis into a long-style flower, the pollen from the stamens stick to the proboscis in exactly the part that will later touch the stigma of the short-styled flower, and vice versa.

Another most important property of the heterostyly system is physiological. If thrum pollen is placed on a thrum stigma, or pin pollen on a pin stigma, the reproductive cells are incompatible and relatively little seed is set. Effectively, this ensures out-crossing, as described by Darwin. Quite a lot is now known about the underlying genetics; the system is controlled by a set of closely linked genes which act as a single unit, a super-gene. All sections of the genus *Primula* have heterostyle species, altogether 354 species out of 419. Since heterostyly is characteristic of nearly all races or species, the system is at least as old as the genus.

Between 1861 and 1863, Darwin found the same kind of structure in other groups: flax (and other species of *Linum*); and in purple loosestrife and other species of *Lythrum*. Some of the *Lythrum* species are trimorphic, with one style and two stamens in each form.

Heterostyly is known in at least 51 genera of 18 families of Angiosperms.

White-throated Sparrows

Black-and-white-striped morph

Brown-and-tan-striped morph

The White-throated Sparrow (*Zonotrichia albicollis*), a passerine bird of the American sparrow family Emberizidae, shows a clear dimorphism in both sexes throughout its large range.

Their heads are either white-striped or tan-striped. These differences in plumage result from a balanced chromosomal inversion polymorphism; in white-striped (WS) birds, one copy of chromosome 2 is partly inverted, while in tan-striped (TS) birds, both copies are uninverted.

The plumage differences are paralleled by differences in behavior and breeding strategy. WS males sing more, are more aggressive and more frequently engage in extra-pair copulation than their TS counterparts. TS birds of both sexes provide more parental care than WS birds.

The polymorphism is maintained by negative assortative mating – each morph mates with its opposite. Dimorphic pairs may have an advantageous balance between parental care and aggressive territorial defense. In addition, as in many other polymorphisms, heterozygote advantage seems to help maintain this one; the proportion of WS birds heterozygotic for the inversion is even lower than would be expected from the low frequency (4%) of pairings of the same morph.

In the underlying chromosomal polymorphism, the standard (ZAL2) and alternative (ZAL2m) arrangements differ by a pair of included pericentric inversions at least. ZAL2m suppresses recombination in the heterokaryotype and is evolving as a rare nonrecombining autosomal segment of the genome.

Darwin's finches

Whereas Darwin spent just five weeks in the Galápagos, and David Lack spent three months, Peter and Rosemary Grant and their colleagues have made research trips to the Galápagos for about thirty years, particularly studying Darwin's finches. Here we look briefly at the case of the large cactus finch *Geospiza conirostris* on Isla Genovesa (formerly Tower Island) which is formed from a shield volcano, and is home to a variety of birds. These birds, like all well-studied groups, show various kinds of morphism.

Males are dimorphic in song type: songs A and B are quite distinct. Also, males with song A have shorter bills than B males. This is also a clear difference. With these beaks males are able to feed differently on their favourite cactus, the prickly pear *Opuntia*. Those with long beaks are able to punch holes in the cactus fruit and eat the fleshy aril pulp which surrounds the seeds, whereas those with shorter beaks tear apart the cactus base and eat the pulp and any insect larvae and pupae (both groups eat flowers and buds). This dimorphism clearly maximises their feeding opportunities during the non-breeding season when food is scarce.

Territories of type A and type B males are random if not mated but alternate if mated: no two breeding males of the same song type shared a common boundary. This initially suggested the possibility of assortative mating by female choice. However, further work showed that "the choice of a male by a female is independent of any conditioning influence of her father's song type {and] there is no evidence of assortative mating by bill type... Hence there is no direct evidence of reproductive subdivision in the population". In 1999 Peter Grant agreed that "sympatric speciation [in this example] is unlikely to occur".

If the population is panmixic, then *Geospiza conirostris* exhibits a balanced genetic polymorphism and not, as originally supposed, a case of nascent sympatric speciation. The selection maintaining the polymorphism maximises the species' niche by expanding its feeding opportunity. The genetics of this situation cannot be clarified in the absence of a detailed breeding program, but two loci with linkage disequilibrium is a possibility.

Another interesting dimorphism is for the bills of young finches, which are either "pink" or "yellow". All species of Darwin's finches exhibit this morphism, which lasts for two months. No interpretation of this phenomenon is known.

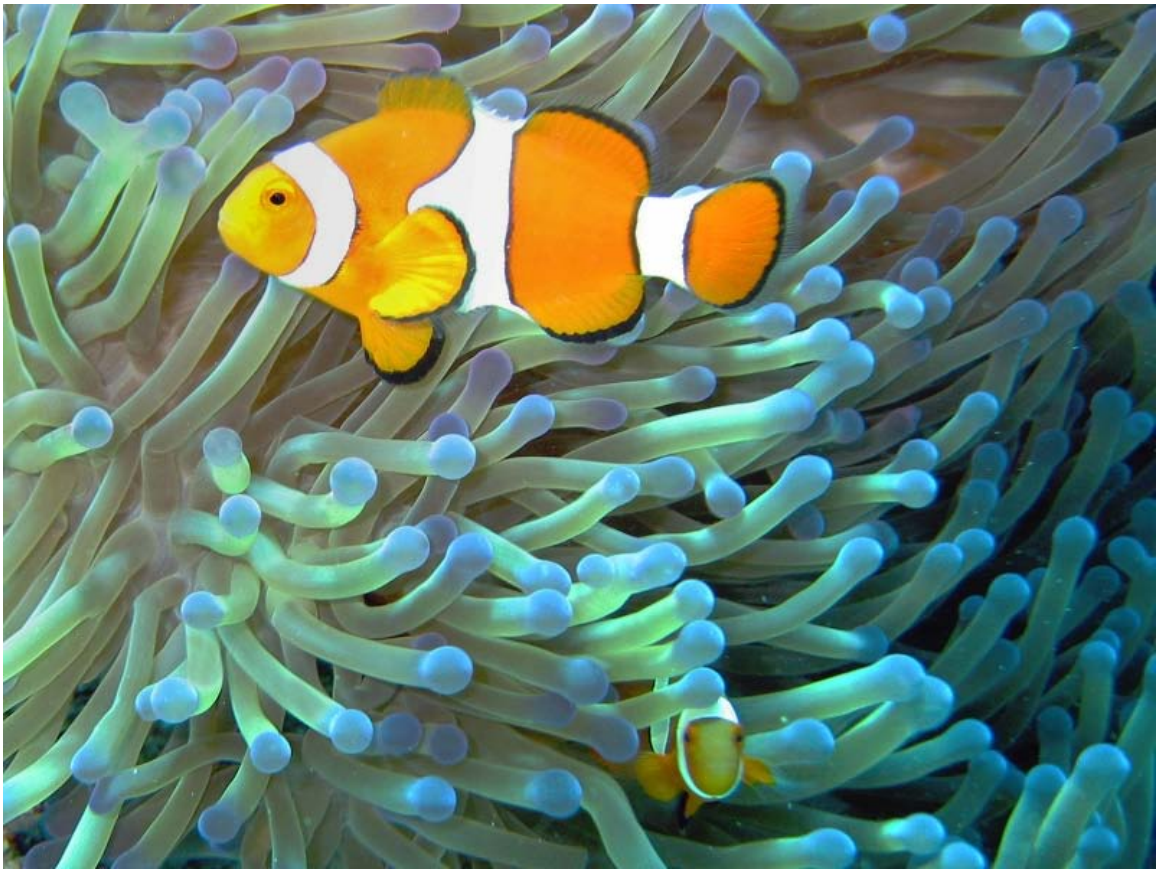
Relative frequency

Endler's survey of natural selection gave an indication of the relative importance of polymorphisms among studies showing natural selection. The results, in summary:

Number of species demonstrating natural selection: 141. Number showing quantitative traits: 56. Number showing polymorphic traits: 62. Number showing both Q and P traits: 23. This shows that polymorphisms are found to be at least as common as continuous variation in studies of natural selection, and hence just as likely to be part of the evolutionary process.

Chapter 3

Symbiosis



In a symbiotic mutualism, the clownfish feeds on small invertebrates which otherwise potentially could harm the sea anemone, and the fecal matter from the clownfish provides nutrients to the sea anemone. The clownfish is additionally protected from predators by the anemone's stinging cells, to which the clownfish is immune.

Symbiosis (from Ancient Greek *sýn* "with" and *bíōsis* "living") is close and often long-term interactions between different biological species. In 1877 Bennett used the word symbiosis (which previously had been used of people living together in community) to

describe the mutualistic relationship in lichens. In 1879 by the German mycologist Heinrich Anton de Bary, defined it as "the living together of unlike organisms."

The definition of symbiosis is in flux, and the term has been applied to a wide range of biological interactions. The symbiotic relationship may be categorized as mutualistic, commensal, or parasitic in nature.

Some symbiotic relationships are obligate, meaning that both symbionts entirely depend on each other for survival. For example, many lichens consist of fungal and photosynthetic symbionts that cannot live on their own. Others are facultative, meaning that they can but do not have to live with the other organism.

Symbiotic relationships include those associations in which one organism lives on another (ectosymbiosis, such as mistletoe), or where one partner lives inside the other (endosymbiosis, such as lactobacilli and other bacteria in humans or zooxanthelles in corals).

Physical interaction

Alder tree root nodule

Endosymbiosis is any symbiotic relationship in which one symbiont lives within the tissues of the other, either in the intracellular space or extracellularly. Examples are

rhizobia, nitrogen-fixing bacteria that live in root nodules on legume roots; actinomycete nitrogen-fixing bacteria called *Frankia*, which live in alder tree root nodules; single-celled algae inside reef-building corals; and bacterial endosymbionts that provide essential nutrients to about 10%–15% of insects.

Ectosymbiosis, also referred to as *exosymbiosis*, is any symbiotic relationship in which the symbiont lives on the body surface of the host, including the inner surface of the digestive tract or the ducts of exocrine glands. Examples of this include ectoparasites such as lice, commensal ectosymbionts such as the barnacles that attach themselves to the jaw of baleen whales, and mutualist ectosymbionts such as cleaner fish.

Mutualism

Hermit crab, *Calcinus laevimanus*, with sea anemone.

Mutualism is any relationship between individuals of different species where both individuals derive a benefit. Generally, only lifelong interactions involving close physical and biochemical contact can properly be considered symbiotic. Mutualistic relationships may be either obligate for both species, obligate for one but facultative for the other, or facultative for both. Many biologists restrict the definition of symbiosis to close mutualist relationships.

A large percentage of herbivores have mutualistic gut fauna that help them digest plant matter, which is more difficult to digest than animal prey. Coral reefs are the result of mutualisms between coral organisms and various types of algae that live inside them. Most land plants and land ecosystems rely on mutualisms between the plants, which fix carbon from the air, and mycorrhyzal fungi, which help in extracting minerals from the ground.

An example of mutual symbiosis is the relationship between the ocellaris clownfish that dwell among the tentacles of Ritteri sea anemones. The territorial fish protects the anemone from anemone-eating fish, and in turn the stinging tentacles of the anemone

protect the clownfish from its predators. A special mucus on the clownfish protects it from the stinging tentacles.

Another example is the goby fish, which sometimes lives together with a shrimp. The shrimp digs and cleans up a burrow in the sand in which both the shrimp and the goby fish live. The shrimp is almost blind, leaving it vulnerable to predators when above ground. In case of danger the goby fish touches the shrimp with its tail to warn it. When that happens both the shrimp and goby fish quickly retract into the burrow.

One of the most spectacular examples of obligate mutualism is between the siboglinid tube worms and symbiotic bacteria that live at hydrothermal vents and cold seeps. The worm has no digestive tract and is wholly reliant on its internal symbionts for nutrition. The bacteria oxidize either hydrogen sulfide or methane which the host supplies to them. These worms were discovered in the late 1980s at the hydrothermal vents near the Galapagos Islands and have since been found at deep-sea hydrothermal vents and cold seeps in all of the world's oceans.

There are also many types of tropical and sub-tropical ants that have evolved very complex relationships with certain tree species.

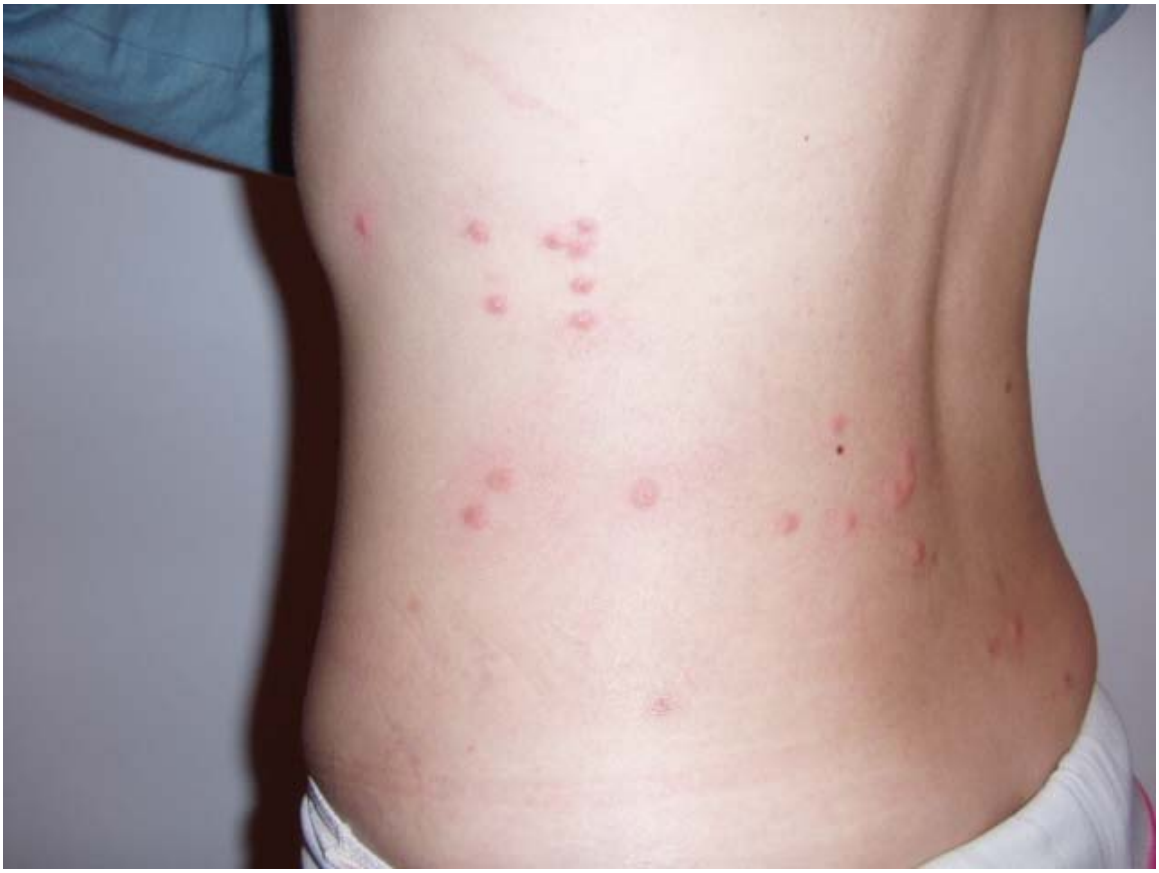
Commensalism

Phoretic mites on a fly (*Pseudolynchia canariensis*).

Commensalism describes a relationship between two living organisms where one benefits and the other is not significantly harmed or helped. It is derived from the English word commensal used of human social interaction. The word derives from the medieval Latin word, formed from *com-* and *mensa*, meaning "sharing a table".

Commensal relationships may involve one organism using another for transportation (phoresy) or for housing (inquilinism), or it may also involve one organism using something another created, after its death (metabiosis). Examples of metabiosis are hermit crabs using gastropod shells to protect their bodies and spiders building their webs on plants.

Parasitism



Flea bites on a human is an example of parasitism (the flea as parasite to the human host in this case).

A parasitic relationship is one in which one member of the association benefits while the other is harmed. Parasitic symbioses take many forms, from endoparasites that live within the host's body to ectoparasites that live on its surface. In addition, parasites may be necrotrophic, which is to say they kill their host, or biotrophic, meaning they rely on their host's surviving. Biotrophic parasitism is an extremely successful mode of life. Depending on the definition used, as many as half of all animals have at least one parasitic phase in their life cycles, and it is also frequent in plants and fungi. Moreover,

almost all free-living animals are host to one or more parasite taxa. An example of a biotrophic relationship would be a tick feeding on the blood of its host.

Amensalism

Amensalism is the type of symbiotic relationship that exists where one species is inhibited or completely obliterated and one is unaffected. This type of symbiosis is relatively uncommon in rudimentary reference texts, but is omnipresent in the natural world. An example is a sapling growing under the shadow of a mature tree. The mature tree can begin to rob the sapling of necessary sunlight and, if the mature tree is very large, it can take up rainwater and deplete soil nutrients. Throughout the process the mature tree is unaffected. Indeed, if the sapling dies, the mature tree gains nutrients from the decaying sapling. Note that these nutrients become available because of the sapling's decomposition, rather than from the living sapling, which would be a case of parasitism.

Symbiosis and evolution

Leafhoppers protected by an army of meat ants

While historically, symbiosis has received less attention than other interactions such as predation or competition, it is increasingly recognised as an important selective force behind evolution, with many species having a long history of interdependent co-evolution. In fact, the evolution of all eukaryotes (plants, animals, fungi, and protists) is believed under the endosymbiotic theory to have resulted from a symbiosis between various sorts of bacteria.

Vascular Plants

Up to 80% of vascular plants worldwide form symbiotic relationships with fungi, for example, in arbuscular mycorrhiza.

Symbiogenesis

The biologist Lynn Margulis, famous for her work on endosymbiosis, contends that symbiosis is a major driving force behind evolution. She considers Darwin's notion of evolution, driven by competition, as incomplete and claims that evolution is strongly based on co-operation, interaction, and mutual dependence among organisms. According to Margulis and Dorion Sagan, "Life did not take over the globe by combat, but by networking."

Co-evolution

Symbiosis played a major role in the co-evolution of flowering plants and the animals that pollinate them. Many plants that are pollinated by insects, bats, or birds have highly specialized flowers modified to promote pollination by a specific pollinator that is also correspondingly adapted. The first flowering plants in the fossil record had relatively simple flowers. Adaptive speciation quickly gave rise to many diverse groups of plants, and, at the same time, corresponding speciation occurred in certain insect groups. Some groups of plants developed nectar and large sticky pollen, while insects evolved more specialized morphologies to access and collect these rich food sources. In some taxa of plants and insects the relationship has become dependent, where the plant species can only be pollinated by one species of insect.

Chapter 4

Polyphenism

Biston betularia caterpillars on birch (left) and willow (right), demonstrating a color polyphenism.

A **polyphenic trait** is a trait for which multiple, discrete phenotypes can arise from a single genotype as a result of differing environmental conditions.

Definition

A **polyphenism** is a biological mechanism that causes a trait to be polyphenic. For example, crocodiles possess a sex-determining polyphenism, and therefore their gender is a polyphenic trait.

When polyphenic forms exist at the same time in the same panmictic (interbreeding) population they can be compared to genetic polymorphism. With polyphenism the switch between morphs is environmental, but with genetic polymorphism with the determination of morph is genetic. These two cases have in common that more than one morph is part of the population at any one time. This is rather different from cases where one morph predictably follows another during, for instance, the course of a year. In essence the latter is normal ontogeny where young forms can and do have different forms, colours and habits to adults.

The discrete nature of polyphenic traits differentiates them from traits like weight and height, which are also dependent on environmental conditions but vary continuously across a spectrum. When a polyphenism is present, an environmental cue causes the organism to develop along a separate pathway, resulting in distinct morphologies; thus, the response to the environmental cue is "all or nothing." The nature of these environmental conditions varies greatly, and includes seasonal cues like temperature and moisture, pheromonal cues, kairomonal cues (signals released from one species that can be recognized by another), and nutritional cues.

Examples of Polyphenism

Sex determination

Sex-determining polyphenisms allow a species to benefit from sexual reproduction while permitting an unequal gender ratio. This can be beneficial to a species because a large female-to-male ratio maximizes reproductive capacity. However, temperature-dependent sex determination (as seen in crocodiles) limits the range in which a species can exist, and makes the species susceptible to endangerment by changes in weather pattern. Temperature-dependent sex determination has been proposed as an explanation for the extinction of the dinosaurs.

Population-dependent and reversible sex determination, found in animals such as the blue wrasse fish, have less potential for failure. In the blue wrasse, only one male is found in a given territory: larvae within the territory develop into females, and adult males will not enter the same territory. If a male dies, one of the females in his territory becomes male, replacing him. While this system ensures that there will always be a mating couple when two animals of the same species are present, it could potentially decrease genetic variance in a population, for example if the females remain in a single male's territory. Furthermore, this system is inherently unstable on a small scale because a single mutation causing a fish to remain permanently male would spread quickly through the population

(due to high female availability) and might eventually cause loss of females in the species, and therefore extinction.

The caste system in insects

The caste system of insects enables eusociality, the division of labor between non-breeding and breeding individuals. A series of polyphenisms determines whether larvae develop into queens, workers, and in some cases soldiers. In the case of the ant, *P. morrisi*, an embryo must develop under certain temperature and photoperiod conditions in order to become a reproductively-active queen. This allows for control of the mating season, but like sex determination, limits the spread of the species into certain climates. In bees, royal jelly provided by worker bees causes a developing larva to become a queen. Royal jelly is only produced when the queen is aging or has died. This system is less subject to influence by environmental conditions, yet prevents unnecessary production of queens.

Seasonal pigmentation changes

Polyphenic pigmentation is adaptive for insect species that undergo multiple mating seasons each year. Different pigmentation patterns provide appropriate camouflage throughout the seasons, as well as alter heat retention as temperatures change. Because insects cease growth and development after eclosion, their pigment pattern is invariable in adulthood: thus, a polyphenic pigment adaptation would be less valuable for species whose adult form survives longer than one year. Birds and mammals, however, are capable of continued physiological changes in adulthood, and some display reversible seasonal polyphenisms, such as coat color in the Arctic fox.

Predator-Induced Polyphenisms

Predator-induced polyphenisms are advantageous because they allow the species to develop in a more reproductively-successful way in a predator's absence, but to otherwise assume a more defensible morphology. However, this advantageous polyphenism can quickly become neutral or a disadvantage if the predator evolves to stop producing the kairomone to which the prey responds. For example, the fly larvae that feed on *Daphnia cucullata* (a water flea) release a kairomone that *Daphnia* can detect. When the fly larvae are present, *Daphnia* grow large helmets that protect them from being eaten. However when the predator is absent, *Daphnia* have smaller heads and are therefore more agile swimmers.

Cannibalistic Polyphenism

The spadefoot toad's polyphenism maximizes its reproductive capacity in temporary desert ponds. While the water is at a safe level, the tadpoles develop slowly on a diet of other opportunistic pond inhabitants. However, when the water level is low and desiccation is imminent, the tadpoles develop a morphology (wide mouth, strong jaw)

that permits them to cannibalize. Cannibalistic tadpoles receive better nutrition and thus metamorphose more quickly, avoiding death as the pond dries up.

Dauer diapause in nematodes

Under conditions of stress such as crowding and high temperature, L1 larvae of some free living nematodes such as C. elegans can switch development to the so called dauer larva state, instead of going the normal molts into a reproductive adult. These dauer larvae are a stress resistant, non-feeding, long-lived stage, enabling the animals to survive harsh conditions. On return to favorable conditions, the animal resumes reproductive development from L3 stage onwards.

Evolution of Polyphenisms

A mechanism has been proposed for the development of polyphenisms:

1. A mutation results in a novel, heritable trait.
2. The trait's frequency expands in the population, creating a population on which selection can act.
3. Pre-existing (background) genetic variation in other genes results in phenotypic differences in expression of the new trait.
4. These phenotypic differences undergo selection; as genotypic differences narrow, the trait becomes:
    1. Genetically fixed (non-responsive to environmental conditions)
    2. Polyphenic (responsive to environmental conditions)

Evolution of novel polyphenisms through this mechanism has been demonstrated in the laboratory. Suzuki and Nijhout used an existing mutation (*black*) in a monophenic green hornworm (*M. sexta*) that causes a black phenotype. They found that if larvae from an existing population of *black* mutants were raised at 20˚C, then all the final instar larvae were black; but if the larvae were instead raised at 28˚C, the final instar larvae ranged in color from black to green. By selecting for larvae that were black if raised at 20˚C but green if raised at 28˚C, they produced a polyphenic strain after thirteen generations.

This fits the model described above because a new mutation (black) was required to reveal pre-existing genetic variation and to permit selection. Furthermore, the production of a polyphenic strain was only possible because of background variation within the species: two alleles, one temperature-sensitive and one stable, were present for a single gene upstream of *black* (in the pigment production pathway) before selection occurred. The temperature-sensitive allele was not observable because at high temperatures, it caused an increase in green pigment in hornworms that were already bright green. However, introduction of the black mutant caused the temperature-dependent changes in pigment production to become obvious. The researchers could then select for larvae with the temperature-sensitive allele, resulting in a polyphenism.

Chapter 5

Mutation

In molecular biology and genetics, **mutations** are changes in a genomic sequence: the DNA sequence of a cell's genome or the DNA or RNA sequence of a virus. They can be defined as sudden and spontaneous changes in the cell. Mutations are caused by radiation, viruses, transposons and mutagenic chemicals, as well as errors that occur during meiosis or DNA replication. They can also be induced by the organism itself, by cellular processes such as hypermutation.

Mutation can result in several different types of change in sequences;(DNA) these can either have no effect, alter the product of a gene, or prevent the gene from functioning properly or completely. Studies in the fly *Drosophila melanogaster* suggest that if a mutation changes a protein produced by a gene, this will probably be harmful, with about 70 percent of these mutations having damaging effects, and the remainder being either neutral or weakly beneficial. Due to the damaging effects that mutations can have on genes, organisms have mechanisms such as DNA repair to remove mutations.

Therefore, the optimal mutation rate for a species is a trade-off between costs of a high mutation rate, such as deleterious mutations, and the metabolic costs of maintaining systems to reduce the mutation rate, such as DNA repair enzymes. Viruses that use RNA as their genetic material have rapid mutation rates, which can be an advantage since these viruses will evolve constantly and rapidly, and thus evade the defensive responses of e.g. the human immune system.

Description

Mutations can involve large sections of DNA becoming duplicated, usually through genetic recombination. These duplications are a major source of raw material for evolving new genes, with tens to hundreds of genes duplicated in animal genomes every million years. Most genes belong to larger families of genes of shared ancestry. Novel genes are produced by several methods, commonly through the duplication and mutation of an ancestral gene, or by recombining parts of different genes to form new combinations with new functions.

Here, domains act as modules, each with a particular and independent function, that can be mixed together to produce genes encoding new proteins with novel properties. For example, the human eye uses four genes to make structures that sense light: three for color vision and one for night vision; all four arose from a single ancestral gene. Another advantage of duplicating a gene (or even an entire genome) is that this increases redundancy; this allows one gene in the pair to acquire a new function while the other copy performs the original function. Other types of mutation occasionally create new genes from previously noncoding DNA.

Changes in chromosome number may involve even larger mutations, where segments of the DNA within chromosomes break and then rearrange. For example, two chromosomes in the *Homo* genus fused to produce human chromosome 2; this fusion did not occur in the lineage of the other apes, and they retain these separate chromosomes. In evolution, the most important role of such chromosomal rearrangements may be to accelerate the divergence of a population into new species by making populations less likely to interbreed, and thereby preserving genetic differences between these populations.

Sequences of DNA that can move about the genome, such as transposons, make up a major fraction of the genetic material of plants and animals, and may have been important in the evolution of genomes. For example, more than a million copies of the Alu sequence are present in the human genome, and these sequences have now been recruited to perform functions such as regulating gene expression. Another effect of these mobile DNA sequences is that when they move within a genome, they can mutate or delete existing genes and thereby produce genetic diversity.

A mutation has caused this garden moss rose to produce flowers of different colors. This is a somatic mutation that may also be passed on in the germ line.

In multicellular organisms with dedicated reproductive cells, mutations can be subdivided into germ line mutations, which can be passed on to descendants through their reproductive cells, and somatic mutations (also called acquired mutations), which involve cells outside the dedicated reproductive group and which are not usually transmitted to descendants. If the organism can reproduce asexually through mechanisms such as cuttings or budding the distinction can become blurred.

For example, plants can sometimes transmit somatic mutations to their descendants asexually or sexually where flower buds develop in somatically mutated parts of plants. A new mutation that was not inherited from either parent is called a *de novo* mutation.

The source of the mutation is unrelated to the consequence, although the consequences are related to which cells were mutated.

Nonlethal mutations accumulate within the gene pool and increase the amount of genetic variation. The abundance of some genetic changes within the gene pool can be reduced by natural selection, while other "more favorable" mutations may accumulate and result in adaptive changes.

For example, a butterfly may produce offspring with new mutations. The majority of these mutations will have no effect; but one might change the color of one of the butterfly's offspring, making it harder (or easier) for predators to see. If this color change is advantageous, the chance of this butterfly surviving and producing its own offspring are a little better, and over time the number of butterflies with this mutation may form a larger percentage of the population.

Neutral mutations are defined as mutations whose effects do not influence the fitness of an individual. These can accumulate over time due to genetic drift. It is believed that the overwhelming majority of mutations have no significant effect on an organism's fitness. Also, DNA repair mechanisms are able to mend most changes before they become permanent mutations, and many organisms have mechanisms for eliminating otherwise permanently mutated somatic cells.

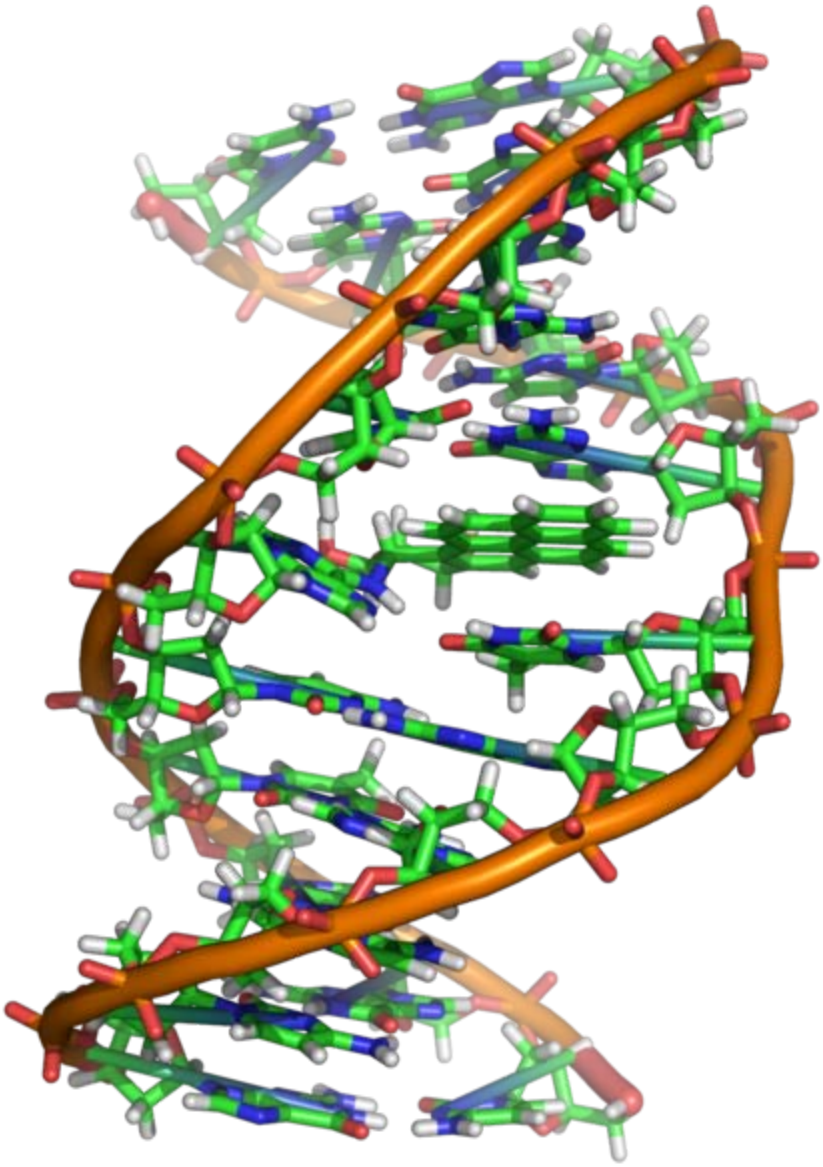
Mutation is generally accepted by biologists as the mechanism by which natural selection acts, generating advantageous new traits that survive and multiply in offspring as well as disadvantageous traits, in less fit offspring, that tend to die out.

Causes

Two classes of mutations are spontaneous mutations (molecular decay) and induced mutations caused by mutagens.

Spontaneous mutations on the molecular level can be caused by:

- Tautomerism – A base is changed by the repositioning of a hydrogen atom, altering the hydrogen bonding pattern of that base resulting in incorrect base pairing during replication.
- Depurination – Loss of a purine base (A or G) to form an apurinic site (AP site).
- Deamination – Hydrolysis changes a normal base to an atypical base containing a keto group in place of the original amine group. Examples include C → U and A → HX (hypoxanthine), which can be corrected by DNA repair mechanisms; and 5MeC (5-methylcytosine) → T, which is less likely to be detected as a mutation because thymine is a normal DNA base.
- Slipped strand mispairing - Denaturation of the new strand from the template during replication, followed by renaturation in a different spot ("slipping"). This can lead to insertions or deletions.



A covalent adduct between benzo[*a*]pyrene, the major mutagen in tobacco smoke, and DNA

Induced mutations on the molecular level can be caused by:

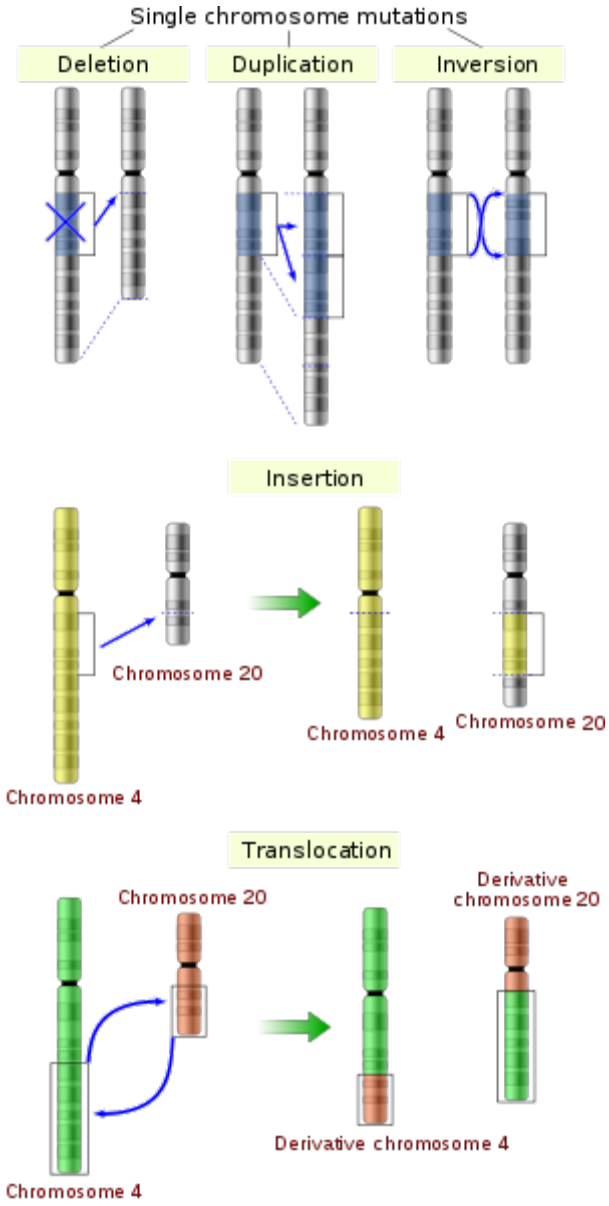
- Chemicals
  - Hydroxylamine NH_2OH
  - Base analogs (e.g. BrdU)
  - Alkylating agents (e.g. *N*-ethyl-*N*-nitrosourea) These agents can mutate both replicating and non-replicating DNA. In contrast, a base analog can only mutate the DNA when the analog is incorporated in replicating the DNA. Each of these classes of chemical mutagens has certain effects that then lead to transitions, transversions, or deletions.

    - Agents that form DNA adducts (e.g. ochratoxin A metabolites)
    - DNA intercalating agents (e.g. ethidium bromide)
    - DNA crosslinkers
    - Oxidative damage
    - Nitrous acid converts amine groups on A and C to diazo groups, altering their hydrogen bonding patterns which leads to incorrect base pairing during replication.
- Radiation
    - Ultraviolet radiation (nonionizing radiation). Two nucleotide bases in DNA – cytosine and thymine – are most vulnerable to radiation that can change their properties. UV light can induce adjacent pyrimidine bases in a DNA strand to become covalently joined as a pyrimidine dimer. UV radiation, particularly longer-wave UVA, can also cause oxidative damage to DNA.
    - Ionizing radiation
    - Radioactive decay, such as ^{14}C in DNA
- Viral infections

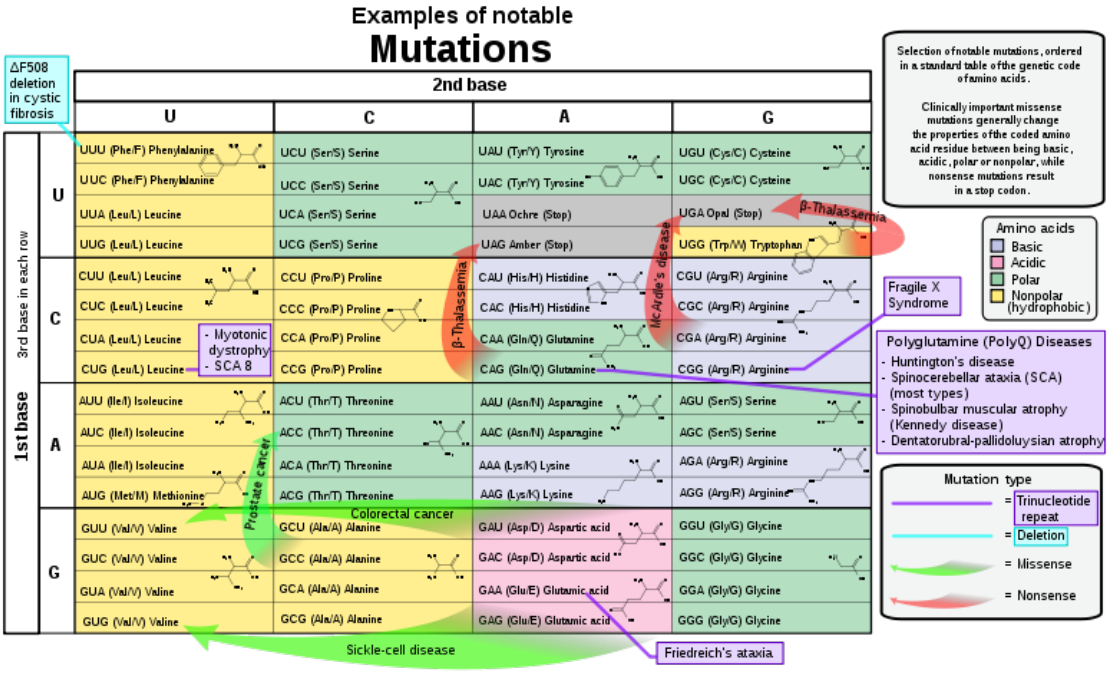
DNA has so-called hotspots, where mutations occur up to 100 times more frequently than the normal mutation rate. A hotspot can be at an unusual base, e.g., 5-methylcytosine.

Mutation rates also vary across species. Evolutionary biologists have theorized that higher mutation rates are beneficial in some situations, because they allow organisms to evolve and therefore adapt more quickly to their environments. For example, repeated exposure of bacteria to antibiotics, and selection of resistant mutants, can result in the selection of bacteria that have a much higher mutation rate than the original population (mutator strains).

Classification of mutation types



Illustrations of five types of chromosomal mutations.



Selection of disease-causing mutations, in a standard table of the genetic code of amino acids.

By effect on structure

The sequence of a gene can be altered in a number of ways. Gene mutations have varying effects on health depending on where they occur and whether they alter the function of essential proteins. Mutations in the structure of genes can be classified as:

- Small-scale mutations, such as those affecting a small gene in one or a few nucleotides, including:
    - **Point mutations**, often caused by chemicals or malfunction of DNA replication, exchange a single nucleotide for another. These changes are classified as transitions or transversions. Most common is the transition that exchanges a purine for a purine (A ↔ G) or a pyrimidine for a pyrimidine, (C ↔ T). A transition can be caused by nitrous acid, base mis-pairing, or mutagenic base analogs such as 5-bromo-2-deoxyuridine (BrdU). Less common is a transversion, which exchanges a purine for a pyrimidine or a pyrimidine for a purine (C/T ↔ A/G). An example of a transversion is adenine (A) being converted into a cytosine (C). A point mutation can be reversed by another point mutation, in which the nucleotide is changed back to its original state (true reversion) or by second-site reversion (a complementary mutation elsewhere that results in regained gene functionality). Point mutations that occur within the protein coding region of a gene may be classified into three kinds, depending upon what the erroneous codon codes for:
        - Silent mutations: which code for the same amino acid.

        - Missense mutations: which code for a different amino acid.
        - Nonsense mutations: which code for a stop and can truncate the protein.
    - **Insertions** add one or more extra nucleotides into the DNA. They are usually caused by transposable elements, or errors during replication of repeating elements (e.g. AT repeats). Insertions in the coding region of a gene may alter splicing of the mRNA (splice site mutation), or cause a shift in the reading frame (frameshift), both of which can significantly alter the gene product. Insertions can be reverted by excision of the transposable element.
    - **Deletions** remove one or more nucleotides from the DNA. Like insertions, these mutations can alter the reading frame of the gene. They are generally irreversible: though exactly the same sequence might theoretically be restored by an insertion, transposable elements able to revert a very short deletion (say 1–2 bases) in *any* location are either highly unlikely to exist or do not exist at all. Note that a deletion is not the exact opposite of an insertion: the former is quite random while the latter consists of a specific sequence inserting at locations that are not entirely random or even quite narrowly defined.
- Large-scale mutations in chromosomal structure, including:
    - **Amplifications** (or gene duplications) leading to multiple copies of all chromosomal regions, increasing the dosage of the genes located within them.
    - **Deletions** of large chromosomal regions, leading to loss of the genes within those regions.
    - Mutations whose effect is to juxtapose previously separate pieces of DNA, potentially bringing together separate genes to form functionally distinct fusion genes (e.g. bcr-abl). These include:
        - **Chromosomal translocations**: interchange of genetic parts from nonhomologous chromosomes.
        - **Interstitial deletions**: an intra-chromosomal deletion that removes a segment of DNA from a single chromosome, thereby apposing previously distant genes. For example, cells isolated from a human astrocytoma, a type of brain tumor, were found to have a chromosomal deletion removing sequences between the "fused in glioblastoma" (fig) gene and the receptor tyrosine kinase "ros", producing a fusion protein (FIG-ROS). The abnormal FIG-ROS fusion protein has constitutively active kinase activity that causes oncogenic transformation (a transformation from normal cells to cancer cells).
        - **Chromosomal inversions**: reversing the orientation of a chromosomal segment.
    - **Loss of heterozygosity**: loss of one allele, either by a deletion or recombination event, in an organism that previously had two different alleles.

By effect on function

- **Loss-of-function mutations** are the result of gene product having less or no function. When the allele has a complete loss of function (null allele) it is often called an **amorphic mutation**. Phenotypes associated with such mutations are most often recessive. Exceptions are when the organism is haploid, or when the reduced dosage of a normal gene product is not enough for a normal phenotype (this is called haploinsufficiency).
- **Gain-of-function mutations** change the gene product such that it gains a new and abnormal function. These mutations usually have dominant phenotypes. Often called a neomorphic mutation.
- **Dominant negative mutations** (also called **antimorphic mutations**) have an altered gene product that acts antagonistically to the wild-type allele. These mutations usually result in an altered molecular function (often inactive) and are characterised by a dominant or semi-dominant phenotype. In humans, Marfan syndrome is an example of a dominant negative mutation occurring in an autosomal dominant disease. In this condition, the defective glycoprotein product of the fibrillin gene (FBN1) antagonizes the product of the normal allele.
- **Lethal mutations** are mutations that lead to the death of the organisms which carry the mutations.
- A **back mutation** or **reversion** is a point mutation that restores the original sequence and hence the original phenotype.

By effect on fitness

In applied genetics it is usual to speak of mutations as either harmful or beneficial.

- A **harmful mutation** is a mutation that decreases the fitness of the organism.
- A **beneficial mutation** is a mutation that increases fitness of the organism, or which promotes traits that are desirable.

In theoretical population genetics, it is more usual to speak of such mutations as deleterious or advantageous. In the neutral theory of molecular evolution, genetic drift is the basis for most variation at the molecular level.

- A **neutral mutation** has no harmful or beneficial effect on the organism. Such mutations occur at a steady rate, forming the basis for the molecular clock.
- A **deleterious mutation** has a negative effect on the phenotype, and thus decreases the fitness of the organism.
- An **advantageous mutation** has a positive effect on the phenotype, and thus increases the fitness of the organism.
- A **nearly neutral mutation** is a mutation that may be slightly deleterious or advantageous, although most nearly neutral mutations are slightly deleterious.

In reality, viewing the fitness effects of mutations in these discrete categories is an oversimplification. Attempts have been made to infer the distribution of fitness effects

using mutagenesis experiments or theoretical models applied to molecular sequence data. However, the current distribution is still uncertain, and some aspects of the distribution likely vary between species.

By inheritance

- inheritable generic in pro-generic tissue or cells on path to be changed to gametes.
- non inheritable **somatic** (e.g., carcinogenic mutation)
- non inheritable post mortem aDNA mutation in decaying remains.

By pattern of inheritance The human genome contains two copies of each gene – a paternal and a maternal allele.

- A **heterozygous mutation** is a mutation of only one allele.
- A **homozygous mutation** is an identical mutation of both the paternal and maternal alleles.
- **Compound heterozygous** mutations or a **genetic compound** comprises two different mutations in the paternal and maternal alleles.
- A **wildtype** or **homozygous non-mutated** organism is one in which neither allele is mutated. (Just not a mutation)

By impact on protein sequence

- A **frameshift mutation** is a mutation caused by insertion or deletion of a number of nucleotides that is not evenly divisible by three from a DNA sequence. Due to the triplet nature of gene expression by codons, the insertion or deletion can disrupt the reading frame, or the grouping of the codons, resulting in a completely different translation from the original. The earlier in the sequence the deletion or insertion occurs, the more altered the protein produced is.

- A **nonsense mutation** is a point mutation in a sequence of DNA that results in a premature stop codon, or a *nonsense codon* in the transcribed mRNA, and possibly a truncated, and often nonfunctional protein product.

- **Missense mutations** or *nonsynonymous mutations* are types of point mutations where a single nucleotide is changed to cause substitution of a different amino acid. This in turn can render the resulting protein nonfunctional. Such mutations are responsible for diseases such as Epidermolysis bullosa, sickle-cell disease, and SOD1 mediated ALS (Boillée 2006, p. 39).

- A **neutral mutation** is a mutation that occurs in an amino acid codon which results in the use of a different, but chemically similar, amino acid. The similarity between the two is enough that little or no change is often rendered in the protein. For example, a change from AAA to AGA will encode arginine, a chemically similar molecule to the intended lysine. Neutral mutations occur because of the degenerate nature of the genetic code.

- **Silent mutations** are mutations that do not result in a change to the amino acid sequence of a protein. They may occur in a region that does not code for a protein, or they may occur within a codon in a manner that does not alter the final amino acid sequence. The phrase *silent mutation* is often used interchangeably with the phrase *synonymous mutation*; however, synonymous mutations are a subcategory of the former, occurring only within exons. The name silent could be a misnomer. For example, a silent mutation in the exon/intron border may lead to alternative splicing by changing the splice site, thereby leading to a changed protein.

Special classes

- **Conditional mutation** is a mutation that has wild-type (or less severe) phenotype under certain "permissive" environmental conditions and a mutant phenotype under certain "restrictive" conditions. For example, a temperature-sensitive mutation can cause cell death at high temperature (restrictive condition), but might have no deleterious consequences at a lower temperature (permissive condition).

Nomenclature

A committee of the Human Genome Variation Society (HGVS) has developed the standard human sequence variant nomenclature, which should be used by researchers and DNA diagnostic centers to generate unambiguous mutation descriptions. In principle, this nomenclature can also be used to describe mutations in other organisms. The nomenclature specifies the type of mutation and base or amino acid changes.

- Nucleotide substitution (e.g. 76A>T) - The number is the position of the nucleotide from the 5' end, the first letter represents the wild type nucleotide, and the second letter represents the nucleotide which replaced the wild type. In the given example, the adenine at the 76th position was replaced by a thymine.
    - If it becomes necessary to differentiate between mutations in genomic DNA, mitochondrial DNA, and RNA, a simple convention is used. For example, if the 100th base of a nucleotide sequence mutated from G to C, then it would be written as g.100G>C if the mutation occurred in genomic DNA, m.100G>C if the mutation occurred in mitochondrial DNA, or r.100g>c if the mutation occurred in RNA. Note that for mutations in RNA, the nucleotide code is written in lower case.
- Amino acid substitution (e.g. D111E) – The first letter is the one letter code of the wild type amino acid, the number is the position of the amino acid from the N terminus, and the second letter is the one letter code of the amino acid present in the mutation. Nonsense mutations are represented with an X for the second amino acid (e.g. D111X).
- Amino acid deletion (e.g. ΔF508) – The Greek letter Δ (delta) indicates a deletion. The letter refers to the amino acid present in the wild type and the number is the position from the N terminus of the amino acid were it to be present as in the wild type.

The complete set of rules and more examples of mutation descriptions can be found at the HGVS sequence variant nomenclature website. Since the nomenclature has to cover all sequence variants, descriptions can become very complex. To prevent mistakes and facilitate correct use of this nomenclature, the journal Human Mutation recommends the use of Mutalyzer, which can apply the HGVS human nomenclature guidelines to check and, if necessary, correct sequence variant descriptions.

Harmful mutations

Changes in DNA caused by mutation can cause errors in protein sequence, creating partially or completely non-functional proteins. To function correctly, each cell depends on thousands of proteins to function in the right places at the right times. When a mutation alters a protein that plays a critical role in the body, a medical condition can result. A condition caused by mutations in one or more genes is called a genetic disorder. Some mutations alter a gene's DNA base sequence but do not change the function of the protein made by the gene. Studies of the fly *Drosophila melanogaster* suggest that if a mutation does change a protein, this will probably be harmful, with about 70 percent of these mutations having damaging effects, and the remainder being either neutral or weakly beneficial. However, studies in yeast have shown that only 7% of mutations that are not in genes are harmful.

If a mutation is present in a germ cell, it can give rise to offspring that carries the mutation in all of its cells. This is the case in hereditary diseases. On the other hand, a mutation may occur in a somatic cell of an organism. Such mutations will be present in all descendants of this cell within the same organism, and certain mutations can cause the cell to become malignant, and thus cause cancer.

Often, gene mutations that could cause a genetic disorder are repaired by the DNA repair system of the cell. Each cell has a number of pathways through which enzymes recognize and repair mistakes in DNA. Because DNA can be damaged or mutated in many ways, the process of DNA repair is an important way in which the body protects itself from disease.

Beneficial mutations

Although most mutations that change protein sequences are neutral or harmful, some mutations have a positive effect on an organism. In this case, the mutation may enable the mutant organism to withstand particular environmental stresses better than wild-type organisms, or reproduce more quickly. In these cases a mutation will tend to become more common in a population through natural selection.

For example, a specific 32 base pair deletion in human CCR5 (CCR5-Δ32) confers HIV resistance to homozygotes and delays AIDS onset in heterozygotes. The CCR5 mutation is more common in those of European descent. One possible explanation of the etiology of the relatively high frequency of CCR5-Δ32 in the European population is that it conferred resistance to the bubonic plague in mid-14th century Europe. People with this

mutation were more likely to survive infection; thus its frequency in the population increased. This theory could explain why this mutation is not found in southern Africa, where the bubonic plague never reached. A newer theory suggests that the selective pressure on the CCR5 Delta 32 mutation was caused by smallpox instead of the bubonic plague.

Another example is Sickle cell disease, a blood disorder in which the body produces an abnormal type of the oxygen-carrying substance hemoglobin in the red blood cells. One-third of all indigenous inhabitants of Sub-Saharan Africa carry the gene, because in areas where malaria is common, there is a survival value in carrying only a single sickle-cell gene (sickle cell trait). Those with only one of the two alleles of the sickle-cell disease are more resistant to malaria, since the infestation of the malaria plasmodium is halted by the sickling of the cells which it infests.

Prion mutation

Prions are proteins and do not contain genetic material. However, prion replication has been shown to be subject to mutation and natural selection just like other forms of replication.

Chapter 6

Abiogenesis

Pre-Cambrian stromatolites in the Siyeh Formation, Glacier National Park. In 2002, William Schopf of UCLA published a paper in the scientific journal *Nature* arguing that geological formations such as this possess 3.5 Ga (billion years old) fossilized cyanobacteria microbes. If true, they would be evidence of the earliest known life on earth.

In natural science, **abiogenesis** is the study of how biological life arises from inorganic matter through natural processes, and the method by which life on Earth arose. Most amino acids, often called "the building blocks of life", can form via natural chemical reactions unrelated to life, as demonstrated in the Miller–Urey experiment and similar experiments that involved simulating some of the conditions of the early Earth in a laboratory. In all living things, these amino acids are organized into proteins, and the

construction of these proteins is mediated by nucleic acids, that are themselves synthesized through biochemical pathways catalysed by proteins. Which of these organic molecules first arose and how they formed the first life is the focus of abiogenesis.

In any theory of abiogenesis, two aspects of life have to be accounted for: replication and metabolism. The question of which came first gave rise to different types of theories. In the beginning, metabolism-first theories (Oparin coacervate) were proposed, and only later thinking gave rise to the modern, replication-first approach.

In modern, still somewhat limited understanding, the first living things on Earth are thought to be single cell prokaryotes (which lack a cell nucleus), perhaps evolved from protobionts (organic molecules surrounded by a membrane-like structure). The oldest ancient fossil microbe-like objects are dated to be 3.5 Ga (billion years old), approximately one billion years after the formation of the Earth itself. By 2.4 Ga, the ratio of stable isotopes of carbon, iron and sulfur shows the action of living things on inorganic minerals and sediments and molecular biomarkers indicate photosynthesis, demonstrating that life on Earth was widespread by this time.

The sequence of chemical events that led to the first nucleic acids is not known. Several hypotheses about early life have been proposed, most notably the iron-sulfur world theory (metabolism without genetics) and the RNA world hypothesis (RNA life-forms).

Conceptual history

Spontaneous generation

Until the early 19th century, people generally believed in the ongoing spontaneous generation of certain forms of life from non-living matter. This was paired with the belief in heterogenesis, e.g. that one form of life derived from a different form (*e.g.* bees from flowers). Classical notions of abiogenesis, now more precisely known as *spontaneous generation*, held that certain complex, living organisms are generated by decaying organic substances. According to Aristotle it was a readily observable truth that aphids arise from the dew which falls on plants, flies from putrid matter, mice from dirty hay, crocodiles from rotting logs at the bottom of bodies of water, and so on.

In the 17th century, such assumptions started to be questioned; for example, in 1646, Sir Thomas Browne published his *Pseudodoxia Epidemica* (subtitled *Enquiries into Very many Received Tenets, and Commonly Presumed Truths*), which was an attack on false beliefs and "vulgar errors." His conclusions were not widely accepted. For example, his contemporary, Alexander Ross wrote: "To question this (i.e., spontaneous generation) is to question reason, sense and experience. If he doubts of this let him go to Egypt, and there he will find the fields swarming with mice, begot of the mud of Nylus, to the great calamity of the inhabitants."

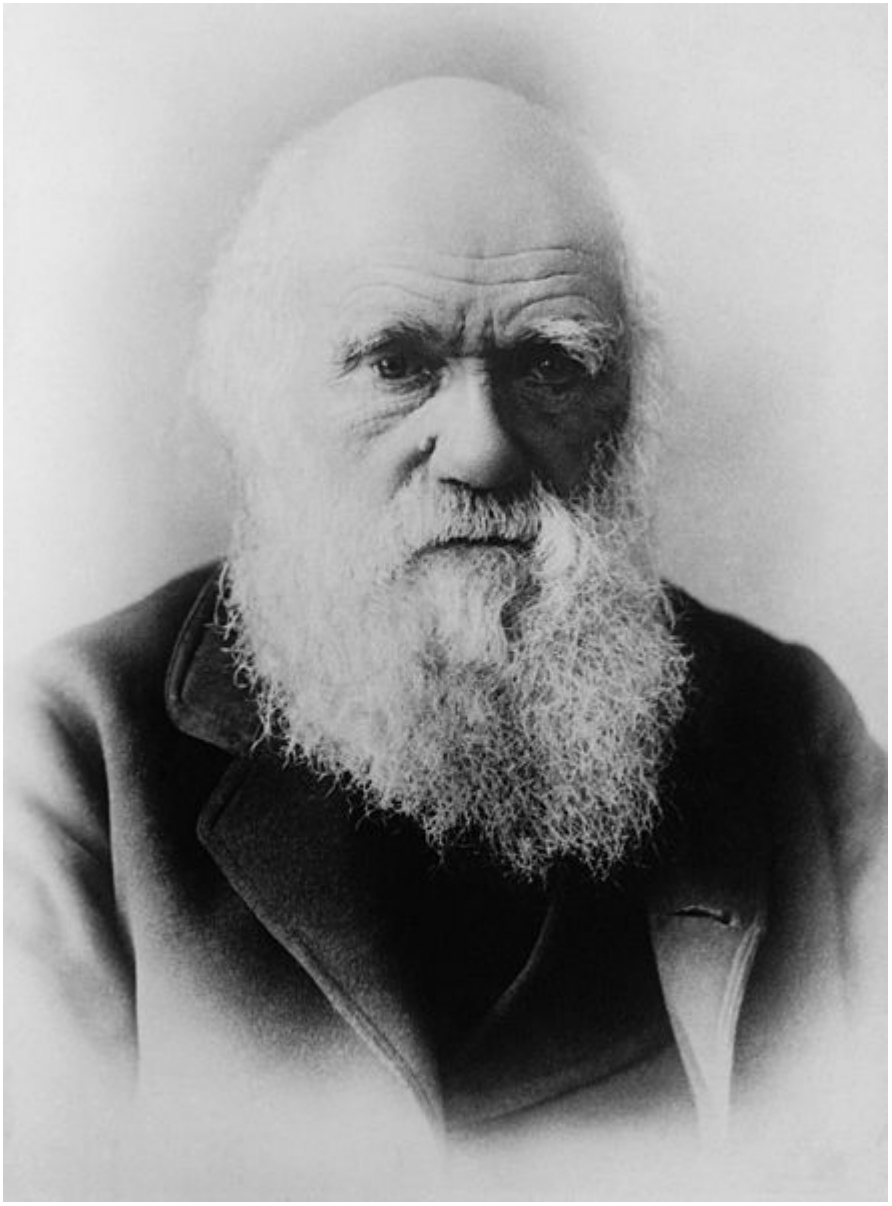
In 1665, Robert Hooke published the first drawings of a microorganism. Hooke was followed in 1676 by Anton van Leeuwenhoek, who drew and described microorganisms

that are now thought to have been protozoa and bacteria. Many felt the existence of microorganisms was evidence in support of spontaneous generation, since microorganisms seemed too simplistic for sexual reproduction, and asexual reproduction through cell division had not yet been observed.

The first solid evidence against spontaneous generation came in 1668 from Francesco Redi, who proved that no maggots appeared in meat when flies were prevented from laying eggs. It was gradually shown that, at least in the case of all the higher and readily visible organisms, the previous sentiment regarding spontaneous generation was false. The alternative seemed to be biogenesis: that every living thing came from a pre-existing living thing (*omne vivum ex ovo*, Latin for "every living thing from an egg").

In 1768, Lazzaro Spallanzani demonstrated that microbes were present in the air, and could be killed by boiling. In 1861, Louis Pasteur performed a series of experiments which demonstrated that organisms such as bacteria and fungi do not spontaneously appear in sterile, nutrient-rich media.

Pasteur and Darwin



Charles Darwin in 1879.

By the middle of the 19th century, the theory of biogenesis had accumulated so much evidential support, due to the work of Louis Pasteur and others, that the alternative theory of spontaneous generation had been effectively disproven. Pasteur himself remarked, after a definitive finding in 1864, "Never will the doctrine of spontaneous generation recover from the mortal blow struck by this simple experiment." The collapse of spontaneous generation, however, left a vacuum of scientific thought on the question of how life *had* first arisen.

In a letter to Joseph Dalton Hooker on February 1, 1871, Charles Darwin addressed the question, suggesting that the original spark of life may have begun in a "warm little pond,

with all sorts of ammonia and phosphoric salts, lights, heat, electricity, etc. present, so that a protein compound was chemically formed ready to undergo still more complex changes". He went on to explain that "at the present day such matter would be instantly devoured or absorbed, which would not have been the case before living creatures were formed." In other words, the presence of life itself makes the search for the origin of life dependent on the sterile conditions of the laboratory.

"Primordial soup" theory

Alexander Oparin (right) at the laboratory.

No new notable research or theory on the subject appeared until 1924, when Alexander Oparin reasoned that atmospheric oxygen prevents the synthesis of certain organic compounds that are necessary building blocks for the evolution of life. In his *The Origin of Life*, Oparin proposed that the "spontaneous generation of life" that had been attacked by Louis Pasteur did in fact occur once, but was now impossible because the conditions found on the early earth had changed, and the presence of living organisms would immediately consume any spontaneously generated organism. Oparin argued that a

"primeval soup" of organic molecules could be created in an oxygenless atmosphere through the action of sunlight. These would combine in evermore complex ways until they formed coacervate droplets. These droplets would "grow" by fusion with other droplets, and "reproduce" through fission into daughter droplets, and so have a primitive metabolism in which those factors which promote "cell integrity" survive, and those that do not become extinct. Many modern theories of the origin of life still take Oparin's ideas as a starting point.

Around the same time, J. B. S. Haldane suggested that the Earth's prebiotic oceans—different from their modern counterparts—would have formed a "hot dilute soup" in which organic compounds could have formed. This idea was called *biopoiesis* or *biopoesis*, the process of living matter evolving from self-replicating but nonliving molecules.

The underlying hypothesis held by Oparin and Haldane was that conditions on the primeval Earth favored chemical reactions that synthesized organic compounds from inorganic precursors. A recent reanalysis of the saved vials containing the original extracts that resulted the Miller and Urey experiments, using current and more advanced analytical equipment and technology, has uncovered more biochemicals than originally discovered in the 1950's. One of the more important findings was 23 amino acids, far more than five originally discovered.

Early conditions

Morse and MacKenzie have suggested that oceans may have appeared first in the Hadean eon, as soon as two hundred million years (200 Ma) after the Earth was formed, in a hot 100 °C (212 °F) reducing environment, and that the pH of about 5.8 rose rapidly towards neutral. This has been supported by Wilde who has pushed the date of the zircon crystals found in the metamorphosed quartzite of Mount Narryer in Western Australia, previously thought to be 4.1–4.2 Ga, to 4.404 Ga. This means that oceans and continental crust existed within 150 Ma of Earth's formation.

Despite this, the Hadean environment was one highly hazardous to life. Frequent collisions with large objects, up to 500 kilometres (310 mi) in diameter, would have been sufficient to vaporise the ocean within a few months of impact, with hot steam mixed with rock vapour leading to high altitude clouds completely covering the planet. After a few months the height of these clouds would have begun to decrease but the cloud base would still have been elevated for about the next thousand years. After that, it would have begun to rain at low altitude. For another two thousand years rains would slowly have drawn down the height of the clouds, returning the oceans to their original depth only 3,000 years after the impact event.

Between 3.8 and 4.1 Ga, changes in the orbits of the gaseous giant planets may have caused a late heavy bombardment that pockmarked the moon and other inner planets (Mercury, Mars, and presumably Earth and Venus). This would likely have sterilized the planet, had life appeared before that time.

By examining the time interval between such devastating environmental events, the time interval when life might first have come into existence can be found for different early environments. The study by Maher and Stevenson shows that if the deep marine hydrothermal setting provides a suitable site for the origin of life, abiogenesis could have happened as early as 4.0 to 4.2 Ga, whereas if it occurred at the surface of the earth abiogenesis could only have occurred between 3.7 and 4.0 Ga.

Other research suggests a colder start to life. Work by Leslie Orgel and colleagues on the synthesis of purines has shown that freezing temperatures are advantageous, due to the concentrating effect for key precursors such as hydrogen cyanide. Research by Stanley Miller and colleagues suggested that while adenine and guanine require freezing conditions for synthesis, cytosine and uracil may require boiling temperatures. Based on this research, Miller suggested a beginning of life involving freezing conditions and exploding meteorites. An article in Discover Magazine points to research by the Miller group indicating the formation of seven different amino acids and 11 types of nucleobases in ice when ammonia and cyanide were left in a freezer from 1972–1997. Here we, also describes research by Christof Biebricher showing the formation of RNA molecules 400 bases long under freezing conditions using an RNA template, a single-strand chain of RNA that guides the formation of a new strand of RNA. As that new RNA strand grows, it adheres to the template. The explanation given for the unusual speed of these reactions at such a low temperature is eutectic freezing. As an ice crystal forms, it stays pure: only molecules of water join the growing crystal, while impurities like salt or cyanide are excluded. These impurities become crowded in microscopic pockets of liquid within the ice, and this crowding causes the molecules to collide more often.

Evidence of the early appearance of life comes from the Isua supercrustal belt in Western Greenland and from similar formations in the nearby Akilia Islands. Carbon entering into rock formations has a ratio of Carbon-13 (^{13}C) to Carbon-12 (^{12}C) of about −5.5 (in units of $\delta^{13}C$), where because of a preferential biotic uptake of ^{12}C, biomass has a $\delta^{13}C$ of between −20 and −30. These isotopic fingerprints are preserved in the sediments, and Mojzis has used this technique to suggest that life existed on the planet already by 3.85 billion years ago. Lazcano and Miller (1994) suggest that the rapidity of the evolution of life is dictated by the rate of recirculating water through mid-ocean submarine vents. Complete recirculation takes 10 million years, thus any organic compounds produced by then would be altered or destroyed by temperatures exceeding 300 °C (572 °F). They estimate that the development of a 100 kilobase genome of a DNA/protein primitive heterotroph into a 7000 gene filamentous cyanobacterium would have required only 7 Ma.

Current models

There is no truly "standard model" of the origin of life. Most currently accepted models draw at least some elements from the framework laid out by the Oparin-Haldane hypothesis. Under that umbrella, however, are a wide array of disparate discoveries and conjectures such as the following, listed in a rough order of postulated emergence:

1. Some theorists suggest that the atmosphere of the early Earth may have been chemically reducing in nature, composed primarily of methane (CH_4), ammonia (NH_3), water (H_2O), hydrogen sulfide (H_2S), carbon dioxide (CO_2) or carbon monoxide (CO), and phosphate (PO_4^{3-}), with molecular oxygen (O_2) and ozone (O_3) either rare or absent.
2. In such a reducing atmosphere, electrical activity can catalyze the creation of certain basic small molecules (monomers) of life, such as amino acids. This was demonstrated in the Miller–Urey experiment by Stanley L. Miller and Harold C. Urey in 1953.
3. Phospholipids (of an appropriate length) can spontaneously form lipid bilayers, a basic component of the cell membrane.
4. A fundamental question is about the nature of the first self-replicating molecule. Since replication is accomplished in modern cells through the cooperative action of proteins and nucleic acids, the major schools of thought about how the process originated can be broadly classified as "proteins first" and "nucleic acids first".
5. The principal thrust of the "nucleic acids first" argument is as follows:
    1. The polymerization of nucleotides into random RNA molecules might have resulted in self-replicating ribozymes (RNA world hypothesis)
    2. Selection pressures for catalytic efficiency and diversity might have resulted in ribozymes which catalyse peptidyl transfer (hence formation of small proteins), since oligopeptides complex with RNA to form better catalysts. The first ribosome might have been created by such a process, resulting in more prevalent protein synthesis.
    3. Synthesized proteins might then outcompete ribozymes in catalytic ability, and therefore become the dominant biopolymer, relegating nucleic acids to their modern use, predominantly as a carrier of genomic information.

No one has synthesized a "protocell" using basic components which would have the necessary properties of life (the so-called *"bottom-up-approach"*). Without such a proof-of-principle, explanations have tended to be short on specifics. However, some researchers are working in this field, notably Steen Rasmussen at Los Alamos National Laboratory and Jack Szostak at Harvard University. Others have argued that a *"top-down approach"* is more feasible. One such approach, successfully attempted by Craig Venter and others at The Institute for Genomic Research, involves engineering existing prokaryotic cells with progressively fewer genes, attempting to discern at which point the most minimal requirements for life were reached. The biologist John Desmond Bernal coined the term *biopoesis* for this process, and suggested that there were a number of clearly defined "stages" that could be recognised in explaining the origin of life.

- Stage 1: The origin of biological monomers
- Stage 2: The origin of biological polymers
- Stage 3: The evolution from molecules to cell

Bernal suggested that evolution may have commenced early, some time between Stage 1 and 2.

Origin of organic molecules

There are two possible sources of organic molecules on the early Earth:

1. Terrestrial origins–organic synthesis driven by impact shocks or by other energy sources (such as ultraviolet light or electrical discharges) (eg.Miller's experiments)
2. Extraterrestrial origins–delivery by objects (e.g. carbonaceous chondrites) or gravitational attraction of organic molecules or primitive life-forms from space

Recently, estimates of these sources suggest that the heavy bombardment before 3.5 Ga within the early atmosphere made available quantities of organics comparable to those produced by other energy sources.

"Soup" theory today: Miller's experiment and subsequent work

Biochemist Robert Shapiro has summarized the "Primordial Soup" theory of Oparin and Haldane in its "mature form" as follows:

1. The early Earth had a chemically reducing atmosphere.
2. This atmosphere, exposed to energy in various forms, produced simple organic compounds ("monomers").
3. These compounds accumulated in a "soup", which may have been concentrated at various locations (Shorelines, oceanic vents etc.).
4. By further transformation, more complex organic polymers— and ultimately life— developed in the soup.

Regarding the reducing atmosphere

Whether the mixture of gases used in the Miller–Urey experiment truly reflects the atmospheric content of early Earth is a controversial topic. Other less reducing gases produce a lower yield and variety. It was once thought that appreciable amounts of molecular oxygen were present in the prebiotic atmosphere, which would have essentially prevented the formation of organic molecules; however, the current scientific consensus is that such was not the case.

Regarding monomer formation

One of the most important pieces of experimental support for the "soup" theory came in 1953. A graduate student, Stanley Miller, and his professor, Harold Urey, performed an experiment that demonstrated how organic molecules could have spontaneously formed from inorganic precursors, under conditions like those posited by the Oparin-Haldane Hypothesis. The now-famous "Miller–Urey experiment" used a highly reduced mixture of gases–methane, ammonia and hydrogen–to form basic organic monomers, such as amino acids. This provided direct experimental support for the second point of the "soup"

theory, and it is around the remaining two points of the theory that much of the debate now centers.

Apart from the Miller–Urey experiment, the next most important step in research on prebiotic organic synthesis was the demonstration by Joan Oró that the nucleic acid purine base, adenine, was formed by heating aqueous ammonium cyanide solutions. In support of abiogenesis in eutectic ice, more recent work demonstrated the formation of s-triazines (alternative nucleobases), pyrimidines (including cytosine and uracil), and adenine from urea solutions subjected to freeze-thaw cycles under a reductive atmosphere (with spark discharges as an energy source).

Regarding monomer accumulation

The "soup" theory relies on the assumption proposed by Darwin that in an environment with no pre-existing life, organic molecules may have accumulated and provided an environment for chemical evolution.

Regarding further transformation

The spontaneous formation of complex polymers from abiotically generated monomers under the conditions posited by the "soup" theory is not at all a straightforward process. Besides the necessary basic organic monomers, compounds that would have prohibited the formation of polymers were formed in high concentration during the Miller–Urey and Oró experiments. The Miller experiment, for example, produces many substances that would undergo cross-reactions with the amino acids or terminate the peptide chain.

More fundamentally, it can be argued that the most crucial challenge unanswered by this theory is how the relatively simple organic building blocks polymerise and form more complex structures, interacting in consistent ways to form a protocell. For example, in an aqueous environment hydrolysis of oligomers/polymers into their constituent monomers would be favored over the condensation of individual monomers into polymers.

The deep sea vent theory

The deep sea vent, or hydrothermal vent, theory for the origin of life on Earth posits that life may have begun at submarine hydrothermal vents, where hydrogen-rich fluids emerge from below the sea floor and interface with carbon dioxide-rich ocean water. Sustained chemical energy in such systems is derived from redox reactions, in which electron donors, such as molecular hydrogen, react with electron acceptors, such as carbon dioxide.

Fox's experiments

In the 1950s and 1960s, Sidney W. Fox studied the spontaneous formation of peptide structures under conditions that might plausibly have existed early in Earth's history. He demonstrated that amino acids could spontaneously form small peptides. These amino

acids and small peptides could be encouraged to form closed spherical membranes, called proteinoid microspheres, which show many of the basic characteristics of 'life'.

Eigen's hypothesis

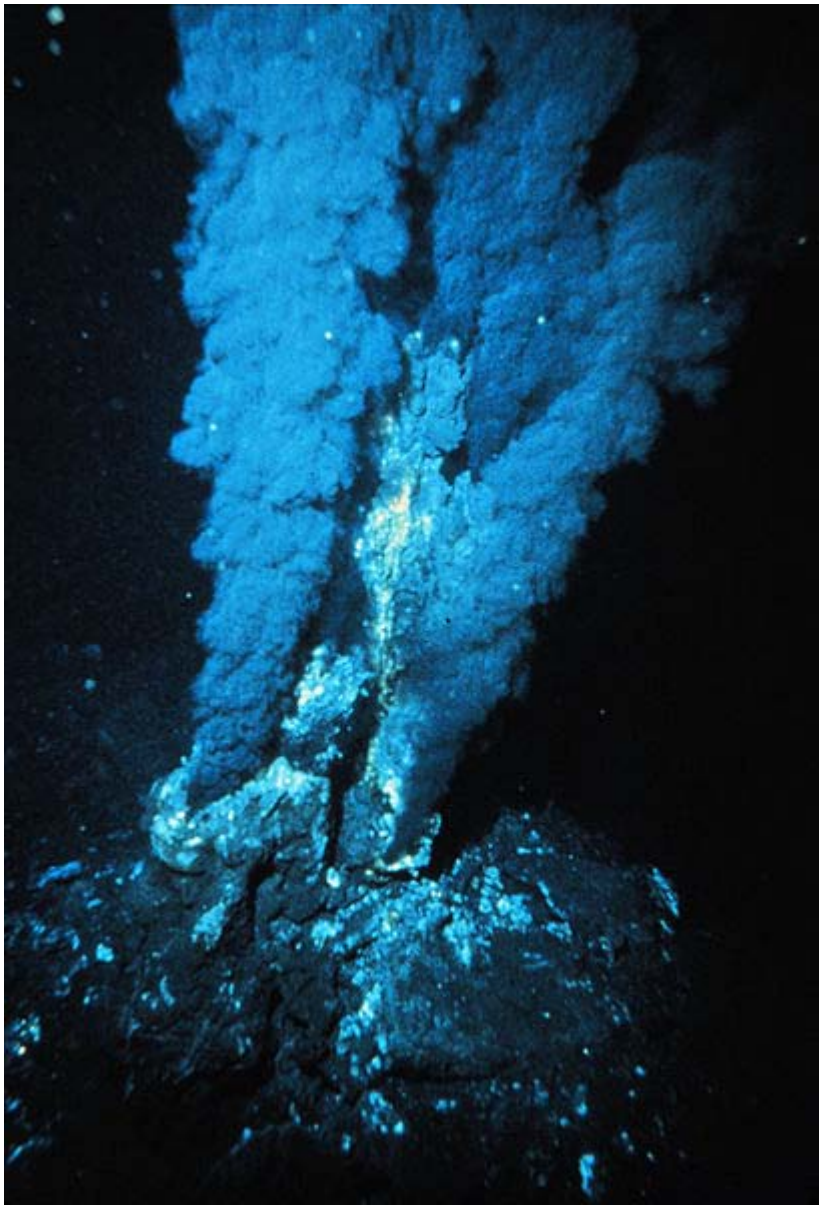
In the early 1970s the problem of the origin of life was approached by Manfred Eigen and Peter Schuster of the Max Planck Institute for Biophysical Chemistry. They examined the transient stages between the molecular chaos and a self-replicating hypercycle in a prebiotic soup.

In a hypercycle, the information storing system (possibly RNA) produces an enzyme, which catalyzes the formation of another information system, in sequence until the product of the last aids in the formation of the first information system. Mathematically treated, hypercycles could create quasispecies, which through natural selection entered into a form of Darwinian evolution. A boost to hypercycle theory was the discovery that RNA, in certain circumstances, forms itself into ribozymes, capable of catalyzing their own chemical reactions. However, these reactions are limited to self-excisions (in which a longer RNA molecule becomes shorter), and much rarer small additions that are incapable of coding for any useful protein. The hypercycle theory is further degraded since the hypothetical RNA would require the existence of complex biochemicals such as nucleotides which are not formed under the conditions proposed by the Miller–Urey experiment.

Hoffmann's contributions

Geoffrey W. Hoffmann, a student of Eigen, contributed to the concept of life involving both replication and metabolism emerging from catalytic noise. His contributions included showing that an early sloppy translation machinery can be stable against an error catastrophe of the type that had been envisaged as problematical by Leslie Orgel ("Orgel's paradox") and calculations regarding the occurrence of a set of required catalytic activities together with the exclusion of catalytic activities that would be disruptive. This is called the stochastic theory of the origin of life.

Wächtershäuser's hypothesis



Deep-sea black smoker

Another possible answer to this polymerization conundrum was provided in 1980s by the German chemist Günter Wächtershäuser, in his iron-sulfur world theory. In this theory, he postulated the evolution of (bio)chemical pathways as fundamentals of the evolution of life. Moreover, he presented a consistent system of tracing today's biochemistry back to ancestral reactions that provide alternative pathways to the synthesis of organic building blocks from simple gaseous compounds.

In contrast to the classical Miller experiments, which depend on external sources of energy (such as simulated lightning or UV irradiation), "Wächtershäuser systems" come

with a built-in source of energy, sulfides of iron and other minerals (e.g. pyrite). The energy released from redox reactions of these metal sulfides is not only available for the synthesis of organic molecules, but also for the formation of oligomers and polymers. It is therefore hypothesized that such systems may be able to evolve into autocatalytic sets of self-replicating, metabolically active entities that would predate the life forms known today.

The experiment produced a relatively small yield of dipeptides (0.4% to 12.4%) and a smaller yield of tripeptides (0.10%) but the authors also noted that: "under these same conditions dipeptides hydrolysed rapidly."

Radioactive beach hypothesis

Zachary Adam at the University of Washington, Seattle, claims that stronger tidal processes from a much closer moon may have concentrated grains of uranium and other radioactive elements at the high water mark on primordial beaches where they may have been responsible for generating life's building blocks. According to computer models reported in *Astrobiology*, a deposit of such radioactive materials could show the same self-sustaining nuclear reaction as that found in the Oklo uranium ore seam in Gabon. Such radioactive beach sand provides sufficient energy to generate organic molecules, such as amino acids and sugars from acetonitrile in water. Radioactive monazite also releases soluble phosphate into regions between sand-grains, making it biologically "accessible". Thus amino acids, sugars and soluble phosphates can all be simultaneously produced, according to Adam. Radioactive actinides, then in greater concentrations, could have formed part of organo-metallic complexes. These complexes could have been important early catalysts to living processes.

John Parnell of the University of Aberdeen suggests that such a process could provide part of the "crucible of life" on any early wet rocky planet, so long as the planet is large enough to have generated a system of plate tectonics which brings radioactive minerals to the surface. As the early Earth is believed to have had many smaller "platelets" it would provide a suitable environment for such processes.

Thermodynamic Origin of Life: Ultraviolet and Temperature-Assisted Replication (UVTAR) Model

Karo Michaelian of the National Autonomous University of Mexico (UNAM) points out that any model for the origin of life must take into account the fact that life is an irreversible thermodynamic process which arises and persists to produce entropy. Entropy production is not incidental to the process of life, but rather the fundamental reason for its existence. Present day life augments the entropy production of Earth by catalysing the water cycle through evapotranspiration. Michaelian argues that if the thermodynamic function of life today is to produce entropy through coupling with the water cycle, then this probably was its function at its very beginnings. It turns out that both RNA and DNA when in water solution are very strong absorbers and extremely rapid dissipaters of ultraviolet light within the 200 nm - 300 nm wavelength range, just

that high energy part of the sun's spectrum that could have penetrated the dense prebiotic atmosphere. Cnossen et al. have shown that the amount of UV light reaching the Earth's surface in the Archean could have been up to 31 orders of magnitude larger than it is today at 260 nm where RNA and DNA absorb most strongly. Absorption and dissipation of UV light by these organic molecules at the Archean ocean surface would have increased significantly the temperature of the surface skin layer leading to enhanced evaporation and thus augmenting the primitive water cycle. Since absorption and dissipation of high energy photons is an entropy producing process, Michaelian argues that non-equilbrium abiogenic synthesis of RNA and DNA utilizing UV light would have been thermodynamically favored.

A simple mechanism to explain the replication of RNA and DNA without the use of enzymes can also be given within the same thermodynamic framework by assuming that life arose when the temperature of the primitive seas had cooled to somewhat below the denaturing temperature of RNA or DNA (based on the ratio of $^{18}O/^{16}O$ found in cherts of the Barberton greenstone belt of South Africa of about 3.5 to 3.2 Ga., surface temperatures are predicted to have been around 70±15 °C, similar to RNA or DNA denaturing temperatures). During the night, the surface water temperature would be below the denaturing temperature and single strand RNA/DNA could act as a template for the formation of double strand RNA/DNA. During the daylight hours, RNA and DNA would absorb UV light and convert this directly to heating of the ocean surface, raising the local temperature enough to allow for denaturing of RNA and DNA. The copying process would be repeated during the cool period overnight. Such a temperature assisted mechanism of replication bears similarity to Polymerase Chain Reaction (PCR), a routine laboratory procedure to multiply DNA segments. Michaelian suggests that traditional origin of life research, expecting to describe the emergence of life from near-equilibrium conditions, is erroneous and that non-equilibrium conditions must be considered, in particular, the importance of entropy production to the emergence of life.

Since denaturation would be most probable in the late afternoon when the Archean sea surface temperature would be highest, and since late afternoon submarine sunlight is somewhat circularly polarized, the homochirality of the organic molecules of life can also be explained within the proposed thermodynamic framework.

Models to explain homochirality

Some process in chemical evolution must account for the origin of homochirality, i.e. all building blocks in living organisms having the same "handedness" (amino acids being left-handed, nucleic acid sugars (ribose and deoxyribose) being right-handed, and chiral phosphoglycerides). Chiral molecules can be synthesized, but in the absence of a chiral source or a chiral catalyst, they are formed in a 50/50 mixture of both enantiomers. This is called a racemic mixture. Clark has suggested that homochirality may have started in space, as the studies of the amino acids on the Murchison meteorite showed L-alanine to be more than twice as frequent as its D form, and L-glutamic acid was more than 3 times prevalent than its D counterpart. It is suggested that polarised light has the power to destroy one enantiomer within the proto-planetary disk. Noyes showed that beta decay

caused the breakdown of D-leucine, in a racemic mixture, and that the presence of ^{14}C, present in larger amounts in organic chemicals in the early Earth environment, could have been the cause. Robert M. Hazen reports upon experiments conducted in which various chiral crystal surfaces act as sites for possible concentration and assembly of chiral monomer units into macromolecules. Once established, chirality would be selected for. Work with organic compounds found on meteorites tends to suggest that chirality is a characteristic of abiogenic synthesis, as amino acids show a left-handed bias, whereas sugars show a predominantly right-handed bias.

Self-organization and replication

While features of self-organization and self-replication are often considered the hallmark of living systems, there are many instances of abiotic molecules exhibiting such characteristics under proper conditions. For example Martin and Russel show that physical compartmentation by cell membranes from the environment and self-organization of self-contained redox reactions are the most conserved attributes of living things, and they argue therefore that inorganic matter with such attributes would be life's most likely last common ancestor.

Virus self-assembly within host cells has implications for the study of the origin of life, as it lends further credence to the hypothesis that life could have started as self-assembling organic molecules.

From organic molecules to protocells

The question "How do simple organic molecules form a protocell?" is largely unanswered but there are many hypotheses. Some of these postulate the early appearance of nucleic acids ("genes-first") whereas others postulate the evolution of biochemical reactions and pathways first ("metabolism-first"). Recently, trends are emerging to create hybrid models that combine aspects of both.

"Genes first" models: the RNA world

The RNA world hypothesis describes an early Earth with self-replicating and catalytic RNA but no DNA or proteins. This has spurred scientists to try to determine if relatively short RNA molecules could have spontaneously formed that were capable of catalyzing their own continuing replication. A number of hypotheses of modes of formation have been put forward. Early cell membranes could have formed spontaneously from proteinoids, protein-like molecules that are produced when amino acid solutions are heated–when present at the correct concentration in aqueous solution, these form microspheres which are observed to behave similarly to membrane-enclosed compartments. Other possibilities include systems of chemical reactions taking place within clay substrates or on the surface of pyrite rocks. Factors supportive of an important role for RNA in early life include its ability to act both to store information and catalyse chemical reactions (as a ribozyme); its many important roles as an intermediate in the expression and maintenance of the genetic information (in the form of DNA) in

modern organisms; and the ease of chemical synthesis of at least the components of the molecule under conditions approximating the early Earth. Relatively short RNA molecules which can duplicate others have been artificially produced in the lab. Such replicase RNA, which functions as both code and catalyst provides a template upon which copying can occur. Jack Szostak has shown that certain catalytic RNAs can, indeed, join smaller RNA sequences together, creating the potential, in the right conditions for self-replication. If these were present, Darwinian selection would favour the proliferation of such self-catalysing structures, to which further functionalities could be added. Lincoln and Joyce identified an RNA enzyme capable of self sustained replication.

Researchers have pointed out difficulties for the abiotic synthesis of nucleotides from cytosine and uracil. Cytosine has a half-life of 19 days at 100 °C (212 °F) and 17,000 years in freezing water. Larralde et al., say that "the generally accepted prebiotic synthesis of ribose, the formose reaction, yields numerous sugars without any selectivity." and they conclude that their "results suggest that the backbone of the first genetic material could not have contained ribose or other sugars because of their instability." The ester linkage of ribose and phosphoric acid in RNA is known to be prone to hydrolysis.

A slightly different version of the RNA-world hypothesis is that a different type of nucleic acid, such as PNA, TNA or GNA, was the first one to emerge as a self-reproducing molecule, to be replaced by RNA only later. Pyrimidine ribonucleosides and their respective nucleotides have been prebiotically synthesised by a sequence of reactions which by-pass the free sugars, and are assembled in a stepwise fashion by going against the dogma that nitrogenous and oxygenous chemistries should be avoided. In a series of publications, The Sutherland Group at the School of Chemistry, University of Manchester have demonstrated high yielding routes to cytidine and uridine ribonucleotides built from small 2 and 3 carbon fragments such as glycolaldehyde, glyceraldehyde or glyceraldehyde-3-phosphate, cyanamide and cyanoacetylene. One of the steps in this sequence allows the isolation of enantiopure ribose aminooxazoline if the enantiomeric excess of glyceraldehyde is 60 % or greater. This can be viewed as a prebiotic purification step, where the said compound spontaneously crystallised out from a mixture of the other pentose aminooxazolines. Ribose aminooxazoline can then react with cyanoacetylene in a mild and highly efficient manner to give the alpha cytidine ribonucleotide. Photoanomerization with UV light allows for inversion about the 1' anomeric centre to give the correct beta stereochemistry. In 2009 they showed that the same simple building blocks allow access, via phosphate controlled nucleobase elaboration, to 2',3'-cyclic pyrimidine nucleotides directly, which are known to be able to polymerise into RNA. This paper also highlights the possibility for the photo-sanitization of the pyrimidine-2',3'-cyclic phosphates. James Ferris's studies have shown that clay minerals of montmorillonite will catalyze the formation of RNA in aqueous solution, by joining activated mono RNA nucleotides to join together to form longer chains. Although these chains have random sequences, the possibility that one sequence began to non-randomly increase its frequency by increasing the speed of its catalysis is possible to "kick start" biochemical evolution.

"Metabolism first" models

Several models reject the idea of the self-replication of a "naked-gene" and postulate the emergence of a primitive metabolism which could provide an environment for the later emergence of RNA replication.

Iron-sulfur world

One of the earliest incarnations of this idea was put forward in 1924 with Alexander Oparin's notion of primitive self-replicating vesicles which predated the discovery of the structure of DNA. More recent variants in the 1980s and 1990s include Günter Wächtershäuser's iron-sulfur world theory and models introduced by Christian de Duve based on the chemistry of thioesters. More abstract and theoretical arguments for the plausibility of the emergence of metabolism without the presence of genes include a mathematical model introduced by Freeman Dyson in the early 1980s and Stuart Kauffman's notion of collectively autocatalytic sets, discussed later in that decade.

However, the idea that a closed metabolic cycle, such as the reductive citric acid cycle, could form spontaneously (proposed by Günter Wächtershäuser) remains debated. In an article entitled "Self-Organizing Biochemical Cycles", the late Leslie Orgel summarized his analysis of the proposal by stating, "There is at present no reason to expect that multistep cycles such as the reductive citric acid cycle will self-organize on the surface of FeS/FeS2 or some other mineral." It is possible that another type of metabolic pathway was used at the beginning of life. For example, instead of the reductive citric acid cycle, the "open" acetyl-CoA pathway (another one of the five recognised ways of carbon dioxide fixation in nature today) would be compatible with the idea of self-organisation on a metal sulfide surface. The key enzyme of this pathway, carbon monoxide dehydrogenase/acetyl-CoA synthase harbours mixed nickel-iron-sulfur clusters in its reaction centers and catalyses the formation of acetyl-CoA (which may be regarded as a modern form of acetyl-thiol) in a single step.

Thermosynthesis world

Today’s bioenergetic process of fermentation is related to the just mentioned citric acid cycle or the Acetyl-CoA pathway that have been connected to the primordial iron-sulfur world. In a different approach, today’s bioenergetic process of chemiosmosis, which plays an essential role in cellular respiration and photosynthesis, is considered as more fundamental than fermentation: in Anthonie Muller’s “thermosynthesis world” the ATP Synthase enzyme that sustains chemiosmosis is proposed as today’s enzyme that is the closest connected to the first metabolic process.

First life needed an energy source to bring about the condensation reaction that yielded the peptide bonds of proteins and the phosphodiester bonds of RNA. In a generalization and thermal variation of the binding change mechanism of today’s ATP Synthase, the “First Protein” would have bound substrates (peptides, phosphate, nucleosides, RNA ‘monomers’) and condensed them to a reaction product that remained bound until it after a temperature change was released upon a thermal unfolding.

The energy source of the thermosynthesis world was thermal cycling, the result of suspension of the protocell in a convection current, as is plausible in a volcanic hot spring; the convection accounts for the self-organization and dissipative structure required in any origin of life model. The still ubiquitous role of thermal cycling in germination and cell division is considered a relic of primordial thermosynthesis.

By phosphorylating cell membrane lipids, this ‘First Protein’ gave a selective advantage to the lipid protocell that contained the protein. In the beginning this First Protein also synthesized a library with many proteins, of which only a minute fraction had thermosynthesis capabilities. Just as proposed by Dyson for the first proteins, the First Protein propagated functionally: it made daughters with similar capabilities, but it did not copy itself. Functioning daughters consisted of different amino acid sequences.

Over a long time, RNA sequences were selected among the at first randomly synthesized RNAs by the criterion of speed and efficiency increase of First Protein synthesis, for instance by the creation of RNA that functioned as messenger RNA, Transfer RNA and ribosomal RNA, or, even more generally, all the components of the RNA World were also generated and selected. The thermosynthesis world therefore in theory accounts for the origin of the genetic machinery.

Whereas the iron-sulfur world identifies a circular pathway as the most simple—and therefore assumes the existence of enzymes—the thermosynthesis world does not even invoke a pathway, and does not assume the existence of regular enzymes: ATP Synthase’s binding change mechanism resembles a physical adsorption process that yields free energy, rather than a regular enzyme’s mechanism, which decreases the free energy. The RNA World also implies the existence of several enzymes. But even the emergence of a single enzyme by chance is implausible. The thermosynthesis world is therefore more simple, and thus more plausible, than the iron-sulfur and RNA worlds.

Possible role of bubbles

Waves breaking on the shore create a delicate foam composed of bubbles. Winds sweeping across the ocean have a tendency to drive things to shore, much like driftwood collecting on the beach. It is possible that organic molecules were concentrated on the shorelines in much the same way. Shallow coastal waters also tend to be warmer, further concentrating the molecules through evaporation. While bubbles composed mostly of water burst quickly, water containing amphiphiles forms much more stable bubbles, lending more time to the particular bubble to perform these crucial reactions.

Amphiphiles are oily compounds containing a hydrophilic head on one or both ends of a hydrophobic molecule. Some amphiphiles have the tendency to spontaneously form membranes in water. A spherically closed membrane contains water and is a hypothetical precursor to the modern cell membrane. If a protein would increase the integrity of its parent bubble, that bubble had an advantage, and was placed at the top of the natural selection waiting list. Primitive reproduction can be envisioned when the bubbles burst, releasing the results of the 'experiment' into the surrounding medium. Once enough of the

'right stuff' was released into the medium, the development of the first prokaryotes, eukaryotes, and multicellular organisms could be achieved.

Similarly, bubbles formed entirely out of protein-like molecules, called microspheres, will form spontaneously under the right conditions. But they are not a likely precursor to the modern cell membrane, as cell membranes are composed primarily of lipid compounds rather than amino-acid compounds.

A recent model by Fernando and Rowe suggests that the enclosure of an autocatalytic non-enzymatic metabolism within protocells may have been one way of avoiding the side-reaction problem that is typical of metabolism first models.

Other models

Autocatalysis

In 1993 Stuart Kauffman proposed that life initially arose as autocatalytic chemical networks.

British ethologist Richard Dawkins wrote about autocatalysis as a potential explanation for the origin of life in his 2004 book *The Ancestor's Tale*. Autocatalysts are substances which catalyze the production of themselves, and therefore have the property of being a simple molecular replicator. In his book, Dawkins cites experiments performed by Julius Rebek and his colleagues at the Scripps Research Institute in California in which they combined amino adenosine and pentafluorophenyl ester with the autocatalyst amino adenosine triacid ester (AATE). One system from the experiment contained variants of AATE which catalysed the synthesis of themselves. This experiment demonstrated the possibility that autocatalysts could exhibit competition within a population of entities with heredity, which could be interpreted as a rudimentary form of natural selection.

Clay theory

A model for the origin of life based on clay was forwarded by A. Graham Cairns-Smith of the University of Glasgow in 1985 and explored as a plausible illustration by several other scientists, including Richard Dawkins. Clay theory postulates that complex organic molecules arose gradually on a pre-existing, non-organic replication platform—silicate crystals in solution. Complexity in companion molecules developed as a function of selection pressures on types of clay crystal is then exapted to serve the replication of organic molecules independently of their silicate "launch stage".

Cairns-Smith is a staunch critic of other models of chemical evolution. However, he admits that like many models of the origin of life, his own also has its shortcomings (Horgan 1991).

In 2007, Kahr and colleagues reported their experiments to examine the idea that crystals can act as a source of transferable information, using crystals of potassium hydrogen

phthalate. "Mother" crystals with imperfections were cleaved and used as seeds to grow "daughter" crystals from solution. They then examined the distribution of imperfections in the crystal system and found that the imperfections in the mother crystals were indeed reproduced in the daughters, but the daughter crystals had many additional imperfections. For gene-like behavior to be observed, the quantity of inheritance of these imperfections should have exceeded that of the mutations in the successive generations, and it did not. Thus Kahr concludes that the crystals "were not faithful enough to store and transfer information from one generation to the next".

Gold's "Deep-hot biosphere" model

In the 1970s, Thomas Gold proposed the theory that life first developed not on the surface of the Earth, but several kilometers below the surface. The discovery in the late 1990s of nanobes (filamental structures that are smaller than bacteria, but that may contain DNA) in deep rocks might be seen as lending support to Gold's theory.

It is now reasonably well established that microbial life is plentiful at shallow depths in the Earth, up to 5 kilometres (3.1 mi) below the surface, in the form of extremophile archaea, rather than the better-known eubacteria (which live in more accessible conditions). It is claimed that discovery of microbial life below the surface of another body in our solar system would lend significant credence to this theory. Thomas Gold also asserted that a trickle of food from a deep, unreachable, source is needed for survival because life arising in a puddle of organic material is likely to consume all of its food and become extinct. Gold's theory is that flow of food is due to out-gassing of primordial methane from the Earth's mantle; more conventional explanations of the food supply of deep microbes (away from sedimentary carbon compounds) is that the organisms subsist on hydrogen released by an interaction between water and (reduced) iron compounds in rocks.

"Primitive" extraterrestrial life

An alternative to Earthly abiogenesis is the hypothesis that primitive life may have originally formed extraterrestrially, either in space or on a nearby planet (Mars). (Note that exogenesis is related to, but not the same as, the notion of panspermia). A supporter of this theory was Francis Crick.

Organic compounds are relatively common in space, especially in the outer solar system where volatiles are not evaporated by solar heating. Comets are encrusted by outer layers of dark material, thought to be a tar-like substance composed of complex organic material formed from simple carbon compounds after reactions initiated mostly by irradiation by ultraviolet light. It is supposed that a rain of material from comets could have brought significant quantities of such complex organic molecules to Earth.

An alternative but related hypothesis, proposed to explain the presence of life on Earth so soon after the planet had cooled down, with apparently very little time for prebiotic evolution, is that life formed first on early Mars. Due to its smaller size Mars cooled

before Earth (a difference of hundreds of millions of years), allowing prebiotic processes there while Earth was still too hot. Life was then transported to the cooled Earth when crustal material was blasted off Mars by asteroid and comet impacts. Mars continued to cool faster and eventually became hostile to the continued evolution or even existence of life (it lost its atmosphere due to low volcanism); Earth is following the same fate as Mars, but at a slower rate.

Neither hypothesis actually answers the question of how life first originated, but merely shifts it to another planet or a comet. However, the advantage of an extraterrestrial origin of primitive life is that life is not required to have evolved on each planet it occurs on, but rather in a single location, and then spread about the galaxy to other star systems via cometary and/or meteorite impact. Evidence to support the hypothesis is scant, but it finds support in recent study of Martian meteorites found in Antarctica and in studies of extremophile microbes. Additional support comes from a recent discovery of a bacterial ecosystem whose energy source is radioactivity.

A 2001 experiment led by Jason Dworkin subjected a frozen mixture of water, methanol, ammonia and carbon monoxide to UV radiation, mimicking conditions found in an extraterrestrial environment. This combination yielded large amounts of organic material that self-organised to form bubbles or micelles when immersed in water. Dworkin considered these bubbles to resemble cell membranes that enclose and concentrate the chemistry of life, separating their interior from the outside world.

The bubbles produced in these experiments were between 10 to 40 micrometres (0.00039 to 0.0016 in), or about the size of red blood cells. Remarkably, the bubbles fluoresced, or glowed, when exposed to UV light. Absorbing UV and converting it into visible light in this way was considered one possible way of providing energy to a primitive cell. If such bubbles played a role in the origin of life, the fluorescence could have been a precursor to primitive photosynthesis. Such fluorescence also provides the benefit of acting as a sunscreen, diffusing any damage that otherwise would be inflicted by UV radiation. Such a protective function would have been vital for life on the early Earth, since the ozone layer, which blocks out the sun's most destructive UV rays, did not form until after photosynthetic life began to produce oxygen.

Extraterrestrial amino acids

Another idea is that amino acids which were formed extra-terrestrially arrived on Earth via comets, consistent with the theory of panspermia. In 2009 it was announced by NASA that scientists have identified one of the fundamental chemical building blocks of life in a comet for the first time: glycine, an amino acid, was detected in the material ejected from Comet Wild-2 in 2004 and grabbed by NASA's Stardust probe. Tiny grains, just a few thousandths of a millimetre in size, were collected from the comet and returned to Earth in 2006 in a sealed capsule, and distributed among the world's leading astro-biology labs. NASA said in a statement that it took some time for the investigating team, led by Dr Jamie Elsila, to convince itself that the glycine signature found in Stardust's sample bay was genuine and not just Earthly contamination. Glycine has been detected in

meteorites before and there are also observations in interstellar gas clouds claimed for telescopes, but the Stardust find is described as a first in cometary material. Isotope analysis indicates that the Late Heavy Bombardment included cometary impacts after the Earth coalesced but before life evolved. Dr. Carl Pilcher, who leads NASA's Astrobiology Institute commented that "The discovery of glycine in a comet supports the idea that the fundamental building blocks of life are prevalent in space, and strengthens the argument that life in the Universe may be common rather than rare."

Lipid World

This theory postulates that the first self-replicating object was lipid-like. It is known that phospholipids form bilayers in water while under agitation– the same structure as in cell membranes. These molecules were not present on early Earth, however other amphiphilic long chain molecules also form membranes. Furthermore, these bodies may expand (by insertion of additional lipids), and under excessive expansion may undergo spontaneous splitting which preserves the same size and composition of lipids in the two progenies. The main idea in this theory is that the molecular composition of the lipid bodies is the preliminary way for information storage, and evolution led to the appearance of polymer entities such as RNA or DNA that may store information favorably. Still, no biochemical mechanism has been offered to support the Lipid World theory.

Polyphosphates

The problem with most scenarios of abiogenesis is that the thermodynamic equilibrium of amino acid versus peptides is in the direction of separate amino acids. What has been missing is some force that drives polymerization. The resolution of this problem may well be in the properties of polyphosphates. Polyphosphates are formed by polymerization of ordinary monophosphate ions PO_4^{-3}. Several mechanisms for such polymerization have been suggested. Polyphosphates cause polymerization of amino acids into peptides. They are also logical precursors in the synthesis of such key biochemical compounds as ATP. A key issue seems to be that calcium reacts with soluble phosphate to form insoluble calcium phosphate (apatite), so some plausible mechanism must be found to keep calcium ions from causing precipitation of phosphate. There has been much work on this topic over the years, but an interesting new idea is that meteorites may have introduced reactive phosphorus species on the early Earth.

PAH world hypothesis

Other sources of complex molecules have been postulated, including extraterrestrial stellar or interstellar origin. For example, from spectral analyses, organic molecules are known to be present in comets and meteorites. In 2004, a team detected traces of polycyclic aromatic hydrocarbons (PAH's) in a nebula. Those are the most complex molecules so far found in space. The use of PAH's has also been proposed as a precursor to the RNA world in the PAH world hypothesis. The Spitzer Space Telescope has recently detected a star, HH 46-IR, which is forming by a process similar to that by which the sun formed. In the disk of material surrounding the star, there is a very large range of

molecules, including cyanide compounds, hydrocarbons, and carbon monoxide. PAHs have also been found all over the surface of galaxy M81, which is 12 million light years away from the Earth, confirming their widespread distribution in space.

Multiple genesis

Different forms of life may have appeared quasi-simultaneously in the early history of Earth. The other forms may be extinct, leaving distinctive fossils through their different biochemistry (e.g., using arsenic instead of phosphorus), survive as extremophiles, or simply be unnoticed through their being analogous to organisms of the current life tree. Hartman for example combines a number of theories together, by proposing that:

The first organisms were self-replicating iron-rich clays which fixed carbon dioxide into oxalic and other dicarboxylic acids. This system of replicating clays and their metabolic phenotype then evolved into the sulfide rich region of the hotspring acquiring the ability to fix nitrogen. Finally phosphate was incorporated into the evolving system which allowed the synthesis of nucleotides and phospholipids. If biosynthesis recapitulates biopoesis, then the synthesis of amino acids preceded the synthesis of the purine and pyrimidine bases. Furthermore the polymerization of the amino acid thioesters into polypeptides preceded the directed polymerization of amino acid esters by polynucleotides.

Lynn Margulis's endosymbiotic theory suggests that multiple forms of archea entered into symbiotic relationship to form the eukaryotic cell. The horizontal transfer of genetic material between archea promotes such symbiotic relationships, and thus many separate organisms may have contributed to building what has been recognised as the Last Universal Common Ancestor (LUCA) of modern organisms. James Lovelock's Gaia theory, proposes that such symbiosis establishes the environment as a system produced by and supportive of life. His arguments strongly weaken the case for life having evolved elsewhere in the solar system.

Chapter 7

Extinction

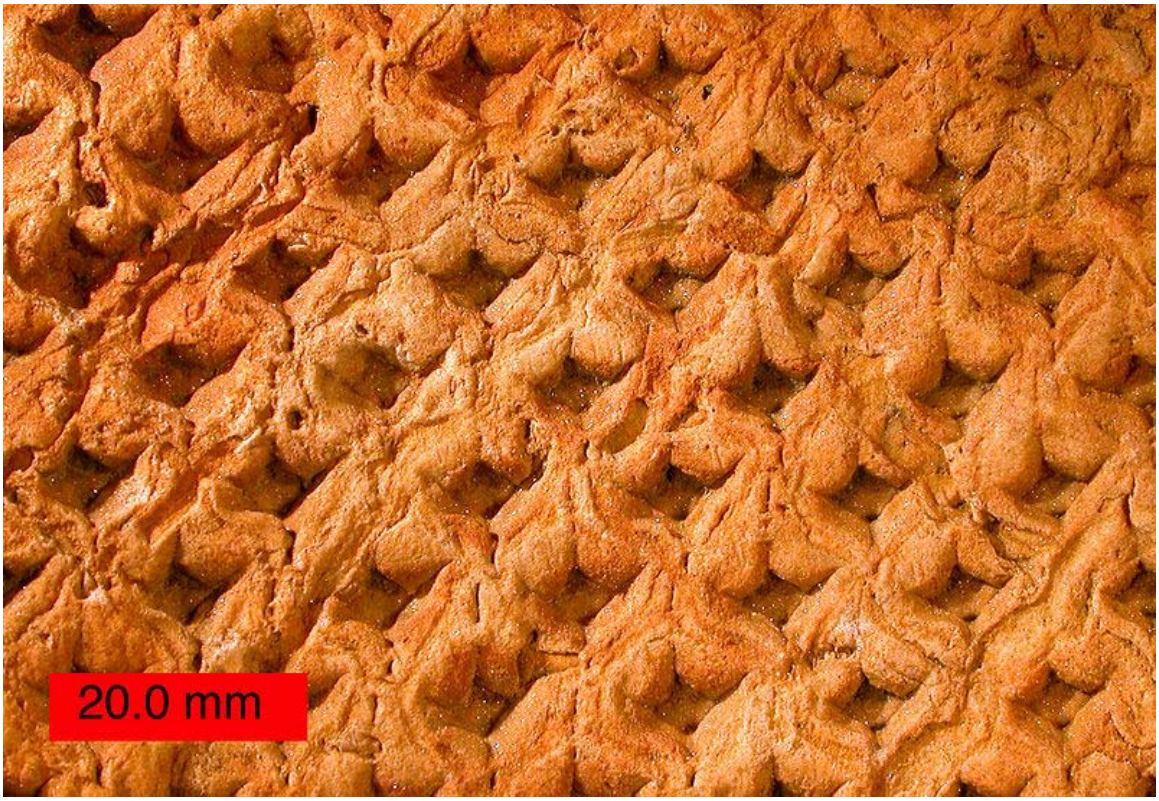
The Dodo, shown here in a 1651 illustration by Jan Savery, is an often-cited example of modern extinction.

In biology and ecology, **extinction** is the end of an organism or of a group of organisms (taxon), normally a species. The moment of extinction is generally considered to be the death of the last individual of the species (although the capacity to breed and recover may have been lost before this point). Because a species' potential range may be very large, determining this moment is difficult, and is usually done retrospectively. This difficulty leads to phenomena such as Lazarus taxa, where a species presumed extinct abruptly "re-appears" (typically in the fossil record) after a period of apparent absence.

Through evolution, new species arise through the process of speciation—where new varieties of organisms arise and thrive when they are able to find and exploit an ecological niche—and species become extinct when they are no longer able to survive in changing conditions or against superior competition. A typical species becomes extinct within 10 million years of its first appearance, although some species, called living fossils, survive virtually unchanged for hundreds of millions of years. Most extinctions occur naturally, without human intervention: it is estimated that 99.9% of all species that have ever existed are now extinct.

Mass extinctions are relatively rare events; however, isolated extinctions are quite common. Only recently have extinctions been recorded and scientists have become alarmed at the high rates of recent extinctions. Most species that become extinct are never scientifically documented. Some scientists estimate that up to half of presently existing species may become extinct by 2100.

Definition



External mold of the extinct *Lepidodendron* from the Upper Carboniferous of Ohio.

A species becomes extinct when the last existing member of that species dies. Extinction therefore becomes a certainty when there are no surviving individuals that are able to reproduce and create a new generation. A species may become functionally extinct when only a handful of individuals survive, which are unable to reproduce due to poor health, age, sparse distribution over a large range, a lack of individuals of both sexes (in sexually reproducing species), or other reasons.

Pinpointing the extinction (or pseudoextinction) of a species requires a clear definition of that species. If it is to be declared extinct, the species in question must be uniquely identifiable from any ancestor or daughter species, or from other closely related species. Extinction of a species (or replacement by a daughter species) plays a key role in the punctuated equilibrium hypothesis of Stephen Jay Gould and Niles Eldredge.

In ecology, *extinction* is often used informally to refer to local extinction, in which a species ceases to exist in the chosen area of study, but still exists elsewhere. This phenomenon is also known as extirpation. Local extinctions may be followed by a replacement of the species taken from other locations; wolf reintroduction is an example of this. Species which are not extinct are termed extant. Those that are extant but threatened by extinction are referred to as threatened or endangered species.

An important aspect of extinction at the present time are human attempts to preserve critically endangered species, which is reflected by the creation of the conservation status "Extinct in the Wild" (EW). Species listed under this status by the International Union for Conservation of Nature (IUCN) are not known to have any living specimens in the wild, and are maintained only in zoos or other artificial environments. Some of these species are functionally extinct, as they are no longer part of their natural habitat and it is unlikely the species will ever be restored to the wild. When possible, modern zoological institutions attempt to maintain a viable population for species preservation and possible future reintroduction to the wild through use of carefully planned breeding programs.

The extinction of one species' wild population can have knock-on effects, causing further extinctions. These are also called "chains of extinction". This is especially common with extinction of keystone species.

Pseudoextinction

Descendants may or may not exist for extinct species. Daughter species that evolve from a parent species carry on most of the parent species' genetic information, and even though the parent species may become extinct, the daughter species lives on. In other cases, species have produced no new variants, or none that are able to survive the parent species' extinction. Extinction of a parent species where daughter species or subspecies are still alive is also called *pseudoextinction*.

Pseudoextinction is difficult to demonstrate unless one has a strong chain of evidence linking a living species to members of a pre-existing species. For example, it is sometimes claimed that the extinct *Hyracotherium*, which was an early horse that shares a common ancestor with the modern horse, is pseudoextinct, rather than extinct, because there are several extant species of *Equus*, including zebra and donkeys. However, as fossil species typically leave no genetic material behind, it is not possible to say whether *Hyracotherium* actually evolved into more modern horse species or simply evolved from a common ancestor with modern horses. Pseudoextinction is much easier to demonstrate for larger taxonomic groups.

Causes

The Passenger Pigeon, one of hundreds of species of extinct birds, was hunted to extinction over the course of a few decades.

The Bali Tiger was declared extinct in 1937 due to hunting and habitat loss.

As long as species have been evolving, species have been going extinct. It is estimated that over 99% of all species that ever lived have gone extinct. The average life-span of most species is 10 million years, although this varies widely between taxa. There are a variety of causes that can contribute directly or indirectly to the extinction of a species or group of species. "Just as each species is unique," write Beverly and Stephen Stearns, "so is each extinction ... the causes for each are varied—some subtle and complex, others obvious and simple". Most simply, any species that is unable to survive or reproduce in its environment, and unable to move to a new environment where it can do so, dies out and becomes extinct. Extinction of a species may come suddenly when an otherwise healthy species is wiped out completely, as when toxic pollution renders its entire habitat unliveable; or may occur gradually over thousands or millions of years, such as when a species gradually loses out in competition for food to better adapted competitors. Extinction may take place a long time after the events that set it in motion, a phenomenon known as extinction debt.

Assessing the relative importance of genetic factors compared to environmental ones as the causes of extinction has been compared to the nature-nurture debate. The question of whether more extinctions in the fossil record have been caused by evolution or by catastrophe is a subject of discussion; Mark Newman, the author of *Modeling Extinction* argues for a mathematical model that falls between the two positions. By contrast, conservation biology uses the extinction vortex model to classify extinctions by cause. When concerns about human extinction have been raised, for example in Sir Martin Rees' 2003 book *Our Final Hour*, those concerns lie with the effects of climate change or technological disaster.

Currently, environmental groups and some governments are concerned with the extinction of species caused by humanity, and are attempting to combat further extinctions through a variety of conservation programs. Humans can cause extinction of a species through overharvesting, pollution, habitat destruction, introduction of new predators and food competitors, overhunting, and other influences. Explosive, unsustainable human population growth is an essential cause of the extinction crisis. According to the International Union for Conservation of Nature (IUCN), 784 extinctions have been recorded since the year 1500 (to the year 2004), the arbitrary date selected to

define "modern" extinctions, with many more likely to have gone unnoticed (several species have also been listed as extinct since the 2004 date).

Genetics and demographic phenomena

Population genetics and demographic phenomena affect the evolution, and therefore the risk of extinction, of species. Limited geographic range is the most important determinant of genus extinction at background rates but becomes increasingly irrelevant as mass extinction arises.

Natural selection acts to propagate beneficial genetic traits and eliminate weaknesses. It is nevertheless possible for a deleterious mutation to be spread throughout a population through the effect of genetic drift.

A diverse or deep gene pool gives a population a higher chance of surviving an adverse change in conditions. Effects that cause or reward a loss in genetic diversity can increase the chances of extinction of a species. Population bottlenecks can dramatically reduce genetic diversity by severely limiting the number of reproducing individuals and make inbreeding more frequent. The founder effect can cause rapid, individual-based speciation and is the most dramatic example of a population bottleneck.

Genetic pollution

Purebred wild species evolved to a specific ecology can be threatened with extinction through the process of genetic pollution—i.e., uncontrolled hybridization, introgression genetic swamping which leads to homogenization or out-competition from the introduced (or hybrid) species. Endemic populations can face such extinctions when new populations are imported or selectively bred by people, or when habitat modification brings previously isolated species into contact. Extinction is likeliest for rare species coming into contact with more abundant ones; interbreeding can swamp the rarer gene pool and create hybrids, depleting the purebred gene pool (for example, the endangered Wild water buffalo is most threatened with extinction by genetic pollution from the abundant domestic water buffalo). Such extinctions are not always apparent from morphological (non-genetic) observations. Some degree of gene flow is a normal evolutionarily process, nevertheless, hybridization (with or without introgression) threatens rare species' existence.

The gene pool of a species or a population is the variety of genetic information in its living members. A large gene pool (extensive genetic diversity) is associated with robust populations that can survive bouts of intense selection. Meanwhile, low genetic diversity reduces the range of adaptions possible. Replacing native with alien genes narrows genetic diversity within the original population, thereby increasing the chance of extinction.

Scorched land resulting from slash-and-burn agriculture.

Habitat degradation

Habitat degradation is currently the main anthropogenic cause of species extinctions. The main cause of habitat degradation worldwide is agriculture, with urban sprawl, logging, mining and some fishing practices close behind. The degradation of a species' habitat may alter the fitness landscape to such an extent that the species is no longer able to survive and becomes extinct. This may occur by direct effects, such as the environment becoming toxic, or indirectly, by limiting a species' ability to compete effectively for diminished resources or against new competitor species.

Habitat degradation through toxicity can kill off a species very rapidly, by killing all living members through contamination or sterilizing them. It can also occur over longer periods at lower toxicity levels by affecting life span, reproductive capacity, or competitiveness.

Habitat degradation can also take the form of a physical destruction of niche habitats. The widespread destruction of tropical rainforests and replacement with open pastureland is widely cited as an example of this; elimination of the dense forest eliminated the infrastructure needed by many species to survive. For example, a fern that depends on dense shade for protection from direct sunlight can no longer survive without forest to shelter it. Another example is the destruction of ocean floors by bottom trawling.

Diminished resources or introduction of new competitor species also often accompany habitat degradation. Global warming has allowed some species to expand their range, bringing unwelcome competition to other species that previously occupied that area. Sometimes these new competitors are predators and directly affect prey species, while at other times they may merely outcompete vulnerable species for limited resources. Vital

resources including water and food can also be limited during habitat degradation, leading to extinction.

The Golden Toad was last seen on May 15, 1989. Decline in amphibian populations is ongoing worldwide.

Predation, competition, and disease

Before the evolution of hominids, life forms competed with each other and drove one another extinct. Recently in geologic time, Humans have been transporting animals and plants from one part of the world to another for thousands of years, sometimes deliberately (e.g., livestock released by sailors onto islands as a source of food) and sometimes accidentally (e.g., rats escaping from boats). In most cases, such introductions are unsuccessful, but when they do become established as an invasive alien species, the consequences can be catastrophic. Invasive alien species can affect native species directly by eating them, competing with them, and introducing pathogens or parasites that sicken or kill them or, indirectly, by destroying or degrading their habitat. Human populations may themselves act as invasive predators. According to the "overkill hypothesis", the swift extinction of the megafauna in areas such as New Zealand, Australia, Madagascar and Hawaii resulted from the sudden introduction of human beings to environments full of animals that had never seen them before, and were therefore completely unadapted to their predation techniques.

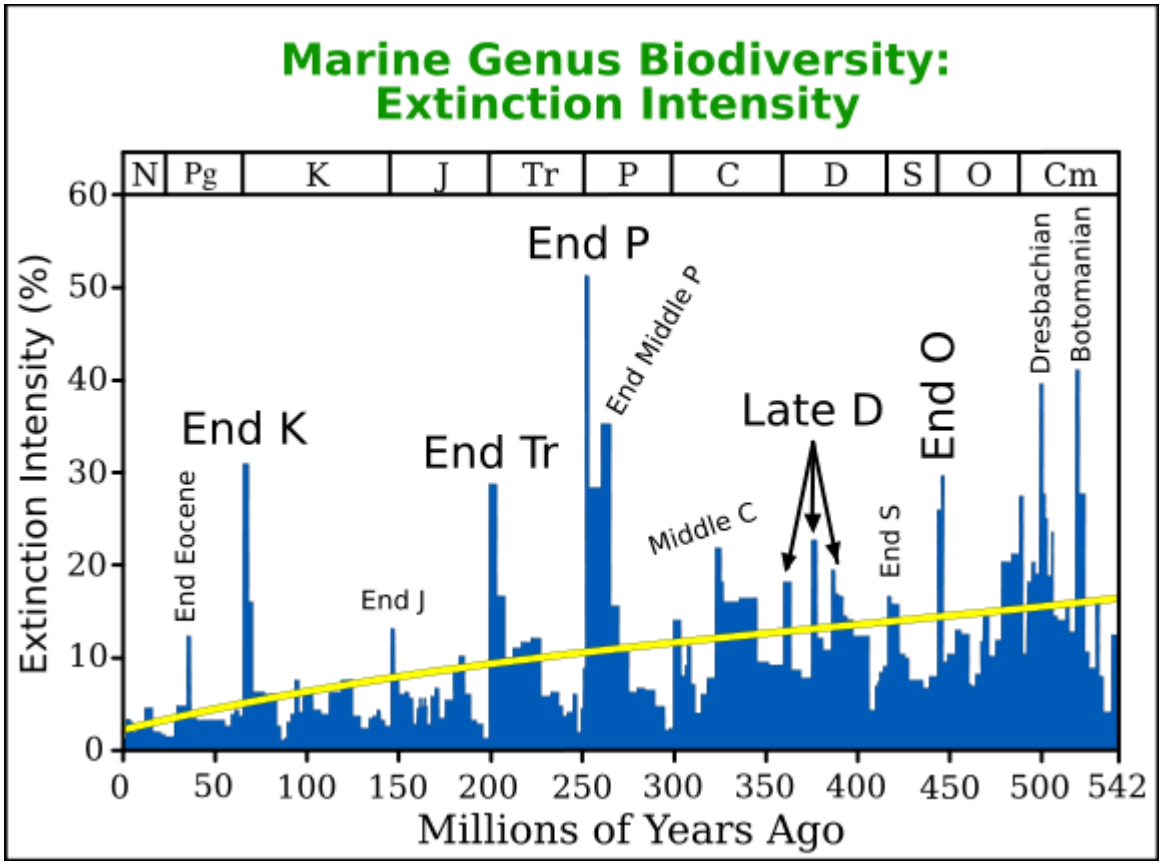
Coextinction

Coextinction refers to the loss of a species due to the extinction of another; for example, the extinction of parasitic insects following the loss of their hosts. Coextinction can also occur when a species loses its pollinator, or to predators in a food chain who lose their prey. "Species coextinction is a manifestation of the interconnectedness of organisms in complex ecosystems ... While coextinction may not be the most important cause of species extinctions, it is certainly an insidious one". Coextinction is especially common when a keystone species goes extinct.

Global warming

A 2003 review across 14 biodiversity research centers predicted that, because of climate change, 15–37% of land species would be "committed to extinction" by 2050. The ecologically rich areas that would potentially suffer the heaviest losses include the Cape Floristic Region, and the Caribbean Basin. These areas might see a doubling of present carbon dioxide levels and rising temperatures that could eliminate 56,000 plant and 3,700 animal species.

Mass extinctions



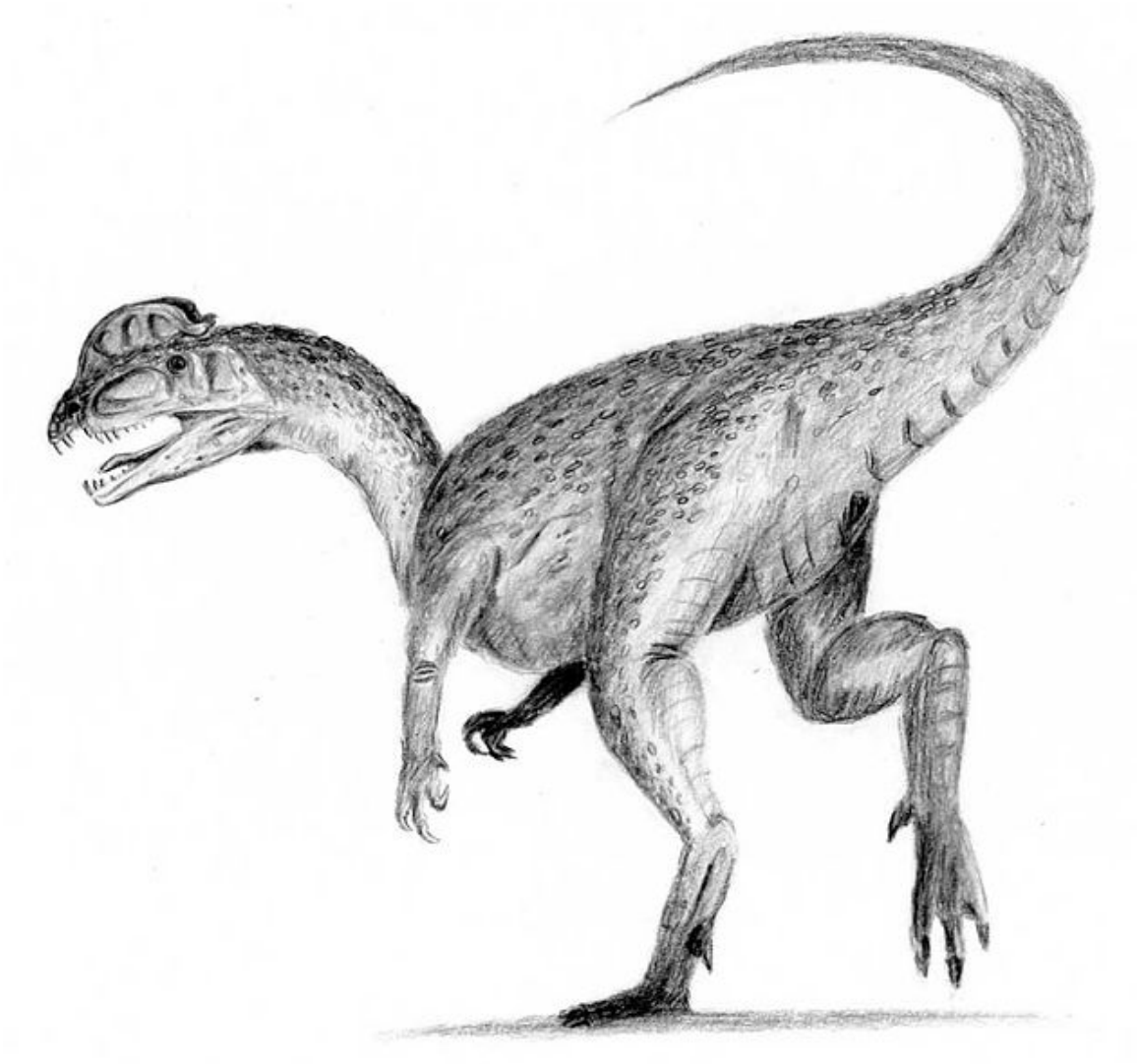
Apparent fraction of genera going extinct at any given time, as reconstructed from the fossil record.

There have been at least five mass extinctions in the history of life on earth, and four in the last 3.5 billion years in which many species have disappeared in a relatively short period of geological time. The most recent of these, the Cretaceous–Tertiary extinction event 65 million years ago at the end of the Cretaceous period, is best known for having wiped out the non-avian dinosaurs, among many other species. The massive eruptive event is considered to be the likely cause of the "Great Dying" about 250 million years ago, which is estimated to have killed 90% of species existing at the time.

Modern extinctions

According to a 1998 survey of 400 biologists conducted by New York's American Museum of Natural History, nearly 70 percent believed that they were currently in the early stages of a human-caused extinction, known as the Holocene extinction. In that survey, the same proportion of respondents agreed with the prediction that up to 20 percent of all living populations could become extinct within 30 years (by 2028). Biologist E. O. Wilson estimated in 2002 that if current rates of human destruction of the biosphere continue, one-half of all species of life on earth will be extinct in 100 years. More significantly the rate of species extinctions at present is estimated at 100 to 1000 times "background" or average extinction rates in the evolutionary time scale of planet Earth.

History of scientific understanding



Dilophosaurus, one of the many extinct dinosaur genera. The cause of the Cretaceous–Tertiary extinction event is a subject of much debate amongst researchers.

In the 1750s when extinction was first described, the idea of extinction was threatening to those who held a belief in the Great Chain of Being, a theological position that did not allow for "missing links".

The possibility of extinction was not widely accepted before the 1800s. The devoted naturalist Carl Linnaeus, could "hardly entertain" the idea that humans could cause the extinction of a species. When parts of the world had not been thoroughly examined and charted, scientists could not rule out that animals found only in the fossil record were not simply "hiding" in unexplored regions of the Earth. Georges Cuvier is credited with establishing extinction as a fact in a 1796 lecture to the French Institute. Cuvier's observations of fossil bones convinced him that they did not originate in extant animals.

This discovery was critical for the spread of uniformitarianism, and lead to the first book publicizing the idea of evolution though Cuvier himself strongly opposed the theories of evolution advanced by Lamarck and others.

Human attitudes and interests

Extinction is an important research topic in the field of zoology, and biology in general, and has also become an area of concern outside the scientific community. A number of organizations, such as the Worldwide Fund for Nature, have been created with the goal of preserving species from extinction. Governments have attempted, through enacting laws, to avoid habitat destruction, agricultural over-harvesting, and pollution. While many human-caused extinctions have been accidental, humans have also engaged in the deliberate destruction of some species, such as dangerous viruses, and the total destruction of other problematic species has been suggested. Other species were deliberately driven to extinction, or nearly so, due to poaching or because they were "undesirable", or to push for other human agendas. One example was the near extinction of the American bison, which was nearly wiped out by mass hunts sanctioned by the United States government, in order to force the removal of Native Americans, many of whom relied on the bison for food.

Biologist Bruce Walsh of the University of Arizona states three reasons for scientific interest in the preservation of species; genetic resources, ecosystem stability, and ethics; and today the scientific community "stress[es] the importance" of maintaining biodiversity.

In modern times, commercial and industrial interests often have to contend with the effects of production on plant and animal life. However, some technologies with minimal, or no, proven harmful effects on *Homo sapiens* can be devastating to wildlife (for example, DDT). Biogeographer Jared Diamond notes that while big business may label environmental concerns as "exaggerated", and often cause "devastating damage", some corporations find it in their interest to adopt good conservation practices, and even engage in preservation efforts that surpass those taken by national parks.

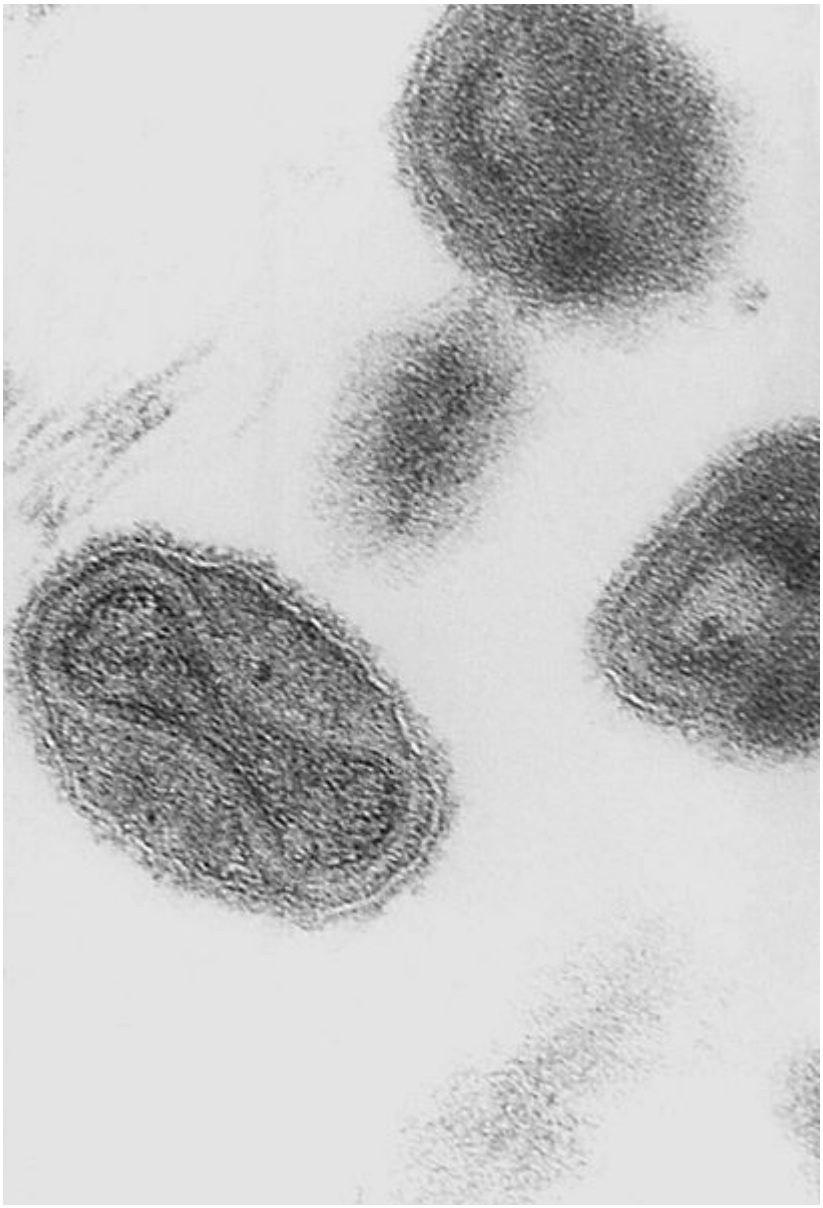
Governments sometimes see the loss of native species as a loss to ecotourism, and can enact laws with severe punishment against the trade in native species in an effort to prevent extinction in the wild. Nature preserves are created by governments as a means to provide continuing habitats to species crowded by human expansion. The 1992 Convention on Biological Diversity has resulted in international Biodiversity Action Plan programmes, which attempt to provide comprehensive guidelines for government biodiversity conservation. Advocacy groups, such as The Wildlands Project and the Alliance for Zero Extinctions, work to educate the public and pressure governments into action.

People who live close to nature can be dependent on the survival of all the species in their environment, leaving them highly exposed to extinction risks. However, people prioritize day-to-day survival over species conservation; with human overpopulation in tropical

developing countries, there has been enormous pressure on forests due to subsistence agriculture, including slash-and-burn agricultural techniques that can reduce endangered species's habitats.

Michael Levin argues, "The very fact that a species is near extinction implies that its final demise will have negligible impact."

Planned extinction



Smallpox virus was eradicated by humans.

Humans have aggressively worked toward the extinction of many species of viruses and bacteria in the cause of disease eradication. For example, the smallpox virus is now extinct in the world—although samples are retained in laboratory settings, and the polio

virus is now confined to small parts of the world as a result of human efforts to prevent the disease it causes.

Olivia Judson is one of six modern scientists to have advocated the deliberate extinction of specific species. Her September 25, 2003 *New York Times* article, "A Bug's Death", advocates "specicide" of thirty mosquito species through the introduction of a genetic element, capable of inserting itself into another crucial gene, to create recessive "knockout genes". Her arguments for doing so are that the *Anopheles* mosquitoes (which spread malaria) and *Aedes* mosquitoes (which spread dengue fever, yellow fever, elephantiasis, and other diseases) represent only 30 species; eradicating these would save at least one million human lives per annum at a cost of reducing the genetic diversity of the family Culicidae by only 1%. She further argues that since species become extinct "all the time" the disappearance of a few more will not destroy the ecosystem: "We're not left with a wasteland every time a species vanishes. Removing one species sometimes causes shifts in the populations of other species — but different need not mean worse." In addition, anti-malarial and mosquito control programs offer little realistic hope to the 300 million people in developing nations who will be infected with acute illnesses this year. Although trials are ongoing, she writes that if they fail: "We should consider the ultimate swatting."

Cloning

Ongoing technological advances have encouraged the hypothesis that by using DNA from the remains of an extinct species, through the process of cloning, the species may be "brought back to life". Proposed targets for cloning include the dinosaurs, the mammoth, thylacine, and the Pyrenean Ibex. In order for such a program to succeed, a sufficient number of individuals would have to be cloned, from the DNA of different individuals (in the case of sexually reproducing organisms) to create a viable population. Though bioethical and philosophical objections have been raised, the cloning of extinct creatures seems a viable outcome of the continuing advancements in our science and technology.

In 2003, scientists attempted to clone the extinct Pyrenean Ibex (*C. p. pyrenaica*). This initial attempt failed; of the 285 embryos reconstructed, 54 were transferred to 12 mountain goats and mountain goat-domestic goat hybrids, but only two survived the initial two months of gestation before they too died. In 2009, a second attempt was made to clone the Pyrenean Ibex; one clone was born alive, but died seven minutes later, due to physical defects in the lungs.

The concept of cloning extinct species was thought to be first popularized by the successful novel and subsequent film *Jurassic Park*, though it may have been first used in John Brosnan's 1984 novel *Carnosaur*, then in F. Paul Wilson's 1989 novel *Dydeetown World,* and later in Piers Anthony's 1990 novel *Balook*, which featured the resurrection of a *Baluchitherium*.