

An introduction to evolution

Biological organization is complex. It ranges from single molecules to highly differentiated cells, to individuals, populations and species interacting in ecological communities. As a consequence biologists asking thousands of seemingly unrelated questions. Why do elephants have trunks? Why do whales have to breath air? Why do viruses lack a metabolism of their own? How can bats navigate during the night? How do proteins assemble? How can plants collect the energy of the sun? Why are salamander genomes 10-times as large as human genomes? Why do humans differ in facial attributes, skin color and behavior? Why are there only about 5,000 mammalian species, but 300,000 of beetles? In one of the most breathtaking ideas in history Charles R. Darwin and Alfred R. Wallace proposed that all living organisms have descended from one primordial form and change through time. This evolutionary concept laid the foundation for a unified theory of biology. Together with insights from the molecular sciences biology transitioned from 'stamp collecting' to a quantitative natural science aiming for a mechanistic and conceptual understanding of life.

Evolution: The process of organismal change and diversification through generational time from shared common ancestry. *Darwin: 'Descent with modification'*.

From this idea it follows that all aspects of biology, the number and arrangement of base pairs in an HIV virus as well as the social organization of ants are the outcome of an evolutionary history that ties all life forms together. The evolutionary perspective thus integrates every subject in biology, from cell biology to ecology. Evolutionary Biology is the unifying theory of biology, or as the influential Russian-American evolutionary geneticist Theodosius Dobzhansky put it: "*Nothing in biology makes sense except in the light of evolution*".

Ernst Mayr introduced an important distinction in this context. Most biological disciplines ask about the **proximate causes** of a phenomenon referring to the mechanistic underpinnings, **how** things work. E.g. the elaborate song in passerine birds is made possible by the syrinx where sound is produced by vibrations of some or all of the *membrana tympaniformis* and the *pessulus* when air is passing through. In addition, evolutionary biology is interested in the **ultimate causes**, the **why** things came into existence in the first place and may persist under current conditions. E.g. male birds sing to claim a territory and attract mating partners, which increases their reproductive success.

Properties of evolution

- Evolution is not the only process generating seemingly **complex forms** (e.g. snowflakes)
- Originally evolution was observed at the level of the phenotype. It is important to recognize that the phenotype is determined by the genotype encoded in DNA (**central dogma**) interacting with the cell and the environment. Ultimately genetic variation is the raw material for evolution.
- Evolution is most readily visible in higher order taxa; yet, it unfolds through *inheritance with modification* of individuals at the population level.
- Evolution does not progress towards a goal, nor is it just random fluctuation.
- Evolution \neq natural selection \neq adaptation.
- Evolution did not just happen in the past. It is ongoing here and now.

Evidence for evolution

Evolution is nothing to believe in, though societal discourse may suggest otherwise in many countries. It is one of the best-supported scientific theories. Evidence for '*descent with modification*' comes from a large number of independent sources.

- **Hierarchical organization of life.** Assuming a historical process of branching and diverging best reflects biological reality of species and higher-order groups with ever decreasing similarity. It is superior to any other organizing system proposed so far.

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- **Homology.** Similarity of structure despite differences in function is best explained by shared ancestry, not by best design.
- **Embryological similarities.** Homologous characters not needed at later life stages appear transiently during embryological development. This can only be understood by shared ancestry.
- **Vestigial characters.** There are numerous examples of ancestral features that are degenerated and of no use to the remnant organism. Cave dwelling fish e.g. display eyes in every stage of degeneration.
- **Convergence.** Functionally similar features often evolved independently and differ profoundly in structure. This provides evidence for adaptation by natural selection and cannot be understood by the hypotheses of optimal design.
- **Suboptimal design.** Indeed, many structures show suboptimal design (e.g. blind spot in human eye) and can only be understood by historical constraint. Evolution simply has to work with the variation present at the time.
- **Geographic distributions.** The distributions of many taxa align well with what we know from the movement of plate tectonics and don't make sense without assuming contiguous change from a common ancestor.
- **Intermediate forms.** The hypothesis of evolution proceeding by gradual small change is well supported by intermediate forms, both in living organisms (some snakes still have vestigial legs) and fossils (e.g. Archaeopteryx).
- **Fossils.** Though incomplete, the fossil record in conjunction with radioactive dating clearly demonstrates that evolution is a long, historical process of change. Dinosaurs are simply not found in the same strata as humans.
- **Molecular evidence.** Common ancestry is seen in a large variety of molecular properties (codon usage, basic molecular machinery, organelles, etc.) and homologies displaying similar functionality across millions of years (if you insert a mouse 'eye' gene into drosophila it will start making an eye providing evidence for a shared bilaterian ancestor).

Evidence for evolution

While scientist from many disciplines still actively investigate the exact mechanisms of evolution today, no compelling evidence has been brought forward against the fact of organismal evolution. When asked what evidence could destroy his confidence in the theory of evolution J.B.S. Haldane answered "fossil rabbits in the pre-cambrian".

Why bother?

Evolutionary Biology has major implications for how we view the world and ourselves. Current day philosophy cannot do without incorporating the findings from molecular and evolutionary biology. It is also of direct applied importance for stock management (e.g. marker assisted breeding), agriculture (e.g. pest control) and the conservation of biodiversity (conservation genetics uses evolutionary principles). Moreover, it is increasingly recognized that evolutionary processes can inform biomedicine in terms of making sense of big data generated to map human disease (e.g. ENCODE project, 1000 human genomes, 23anMe, deCODE), understanding disease (e.g. evolutionary medicine, modeling cancer with population genetic methods, recognition of HIV as evolving zoonosis) and therapy (e.g. mitochondrial replacement therapy, antibiotics resistance). Evolutionary biology has even made its way into industrial production, where processes and products are optimized using evolutionary algorithms.

Historical background

Historical awareness is crucial to understand current debates and to question current day beliefs.

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before Darwin.

Ideas of how biodiversity had originated and was organized were fundamentally different before Darwin entered the historical stage. Greek philosophy had substantial influence on the western tradition of perceiving the world. The point of departure is metaphysical and reaches well into the 19th century and still influences many debates today.

Plato (428-348 BC) the world is a shadow of underlying essences in a metaphysical world; all variation we see in the world simply reflects an imperfect earthly materialization of the essences that are preconfigured in the **metaphysical** world. This view is incompatible with species transformation or evolution.

Aristotle (384-322 BC) centers his view more on the real world and emphasizes experimentation. He recognized change during an individual's lifetime and conceptualizes evo-devo by contrasting two possible mechanisms for reproduction and development: **epigenesis** (embryological differentiation from homogenous material) and **preformation** (development simply entails a preprogrammed unfolding); yet species are fixed and aligned in ladder of life (*Scala Naturae*) from (seemingly) least to most complex (humans of course).

During the **Middle ages** Greek philosophers remain highly influential, the *Scala Naturae* is modified into the **Great Chain of Being**, adding angels, demons and God. *Natural theology* develops with the motivation to reveal the wonders of creation by the study of nature. Thomas Aquinas (1225-1274) describes perfection and adaptation as evidence for divine design (which still prevails in the mind of many creationists world wide). Christianity also introduced the concept that **creation was recent**. Bishop Ussher (1581-1656) went through the biblical genealogies and concluded that creation occurred 4004 BC (nightfall of 22nd October).

In the 18th century the Swedish lecturer **Carolus Linnaeus** develops a modern **biological nomenclature** using a binomial system with species being grouped in genera and higher order taxa to organize biodiversity in his *Systema Naturae* (1735). While this nomenclature inherently recognizes differences in similarity between species and thus implies common ancestry, Linnaeus doesn't make this step. The system is still thought to reflect God's plan. All variation on earth is not of interest and, as for Plato, simply reflects imperfection - deviation from a divine model.

In the late 18th century doubt arises as to the recent age of the earth. Methods for dating rock strata (Georges Cuvier) and evidence for marine fossils high up on inland mountains suggest that the earth is bound to be older than 6000 years. Fossil excavations are booming and provide clear evidence for both speciation and extinction. Geologists such as Charles Lyell and James Hutton (called **uniformitarians**) proposed that geological change was best explained by gradual changes over a very long time span by the same forces that can be observed today (erosion, volcanism). These ideas were highly influential to their coeval Charles Darwin.

The 18th century is also the age of colonialism. Expeditions across the globe are organized to exploit new crop species, a precondition for Darwin's **voyage of the Beagle** where his ideas materialize. Darwin was not the first to question constancy of species and conceptualize evolution. Evolution was 'in the air' at the time. In 1844, e.g. Robert Chambers published a book in which he describes organisms to arise by spontaneous generation and progressively transform into ever more complex forms. These ideas are similar to those of **Jean-Baptiste Lamarck** who also argued that species are transformed and change according to the inheritance of acquired characters. The idea of trans-generational inheritance without genetic change has seen an interesting revival today in the growing field of epigenetics.

after Charles Darwin

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In contrast to Lamarck, Darwin recognized that evolution does not have a goal. He introduced natural selection as a principle for evolutionary change. Influenced by the economic theoretician **Thomas R. Malthus** he realized that many more offspring are produced than can make it into the next generation (overproduction). Together with **Alfred Russel Wallace**, a professional collector working the East Indies who had conceived of the same principle, he published this idea back-to-back in 1858. A year later, his seminal book 'On the Origin of Species by Means of Natural Selection or the Preservation of Favoured Races in the Struggle for Life' is published providing overwhelming evidence that he had amassed over many years.

The basic ingredients of Darwin's theory are:

- heritable trait variation
- overproduction (but fixed resources leading to selection)
- variation in lifetime reproductive success linked to variation in the trait

His contemporaries immediately accepted his theory of descent with modification. However, the mechanism of natural selection remained controversial, also due to his incapacity of explaining the **hereditary basis for the variation** selection can act on. He could have found the answer in the work of his coeval **Gregor Mendel**. By 1866, this Moravian monk had established the essentials of our modern understanding of inheritance (*segregation, independent assortment, dominance*) by meticulous crossing of pea varieties (and counting about 350,000 seeds!). Mendel showed that a pair of elements, one maternal and one paternal, determined the phenotype of the offspring. Yet, at the time few seemed interested in the study of inheritance per se, and the work was only rediscovered in 1900. Hugo de Vries, Carl Correns and Erich Tschermak von Seysenegg independently came to the same conclusions as Mendel - only to discover they were 35 years late.

This rediscovery, however, had a paradoxical effect. It wasn't taken as evidence for Darwin's gradualism, but instead deepened the divide between saltationists who emphasized the importance of discontinuous variation (evolutionary sports) and Darwinian gradualists. It took another half century until Darwinian evolution and Mendelian principles of inheritance were fully reconciled primarily by work of **John B. S. Haldane, Ronald A. Fisher and Sewall Wright** in what we now refer to as the **Modern Evolutionary Synthesis**. Building on previous work including that by **Nielson Ehle**, who showed that continuous variation can be encoded by discrete genetic factors, the Evolutionary Synthesis paved the way for a firm mathematical evolutionary framework of evolution. It established that evolution progresses by micro-evolutionary principles acting on genetically encoded variation. This Synthesis, which was took place in the 1940ies provided an explanatory for hithero unconnected disciplines ranging from paleontology, botany, zoology, experimental biology and natural history.

Genetic variation and the evolutionary forces generating (mutation) and depleting it (selection, genetic drift) lie at the center of the Synthesis. In the absence of empirical data quantifying the amount of genetic variation, a heated debated starts on the nature of genetic variation, as to how it arises and what maintains it. The **classical view** with **Thomas H. Morgan** and **Herman Muller** as its prominent proponents holds that a single (wild-type) form has highest fitness and all other variants are purged by selection. As a consequence genetic variation in natural populations should be very low. The **balancing school** on the contrary (Dobzhansky) assumes a creative role for selection stating that different alleles are maintained by **balancing selection** – providing ample raw material for adaptive evolution. The debated required empirical input.

This input was provided by **Richard Lewontin**, who just won the Craaford Price. Together with Hubby he demonstrated with **protein electrophoresis** that the amount of genetic variation was even higher than previously thought, even higher than proposed by the balance

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school. The same result was later shown for DNA sequence data, who had just been discovered to be the blueprint of all replicating genetic information (**Rosalind Franklin, James D. Watson, Francis H. C. Crick, 1953**).

The first observations of molecular diversity and divergence prompted **Motoo Kimura** 1968 to formulate a **Neutral Theory of Evolution**. This theory states that “the great majority of evolutionary changes at the molecular level (...) are caused not by Darwinian selection, but by random drift of selectively neutral or nearly neutral variants”. It puts a premium on mutation and random drift as the main explanatory factors for both polymorphism and divergence. Selection only plays a role for removing strongly deleterious variants. A very small proportion are advantageous and quickly brought to fixation. The vast majority of the polymorphisms we can observe are selectively neutral. The Neutral Theory establishes the conceptual foundation of the molecular clock, as molecular evolution is assumed to equal neutral mutation rate. **Tomoko Ohta** refined the Neutral Theory (**Nearly Neutral Theory**) allowing for mutations with continuous fitness effects (not only with dichotomous effects neutral or so strong negative that purged immediately); slightly deleterious (advantageous) mutations also possible. This addition has the important consequence that the efficacy of selection depends on population size.

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