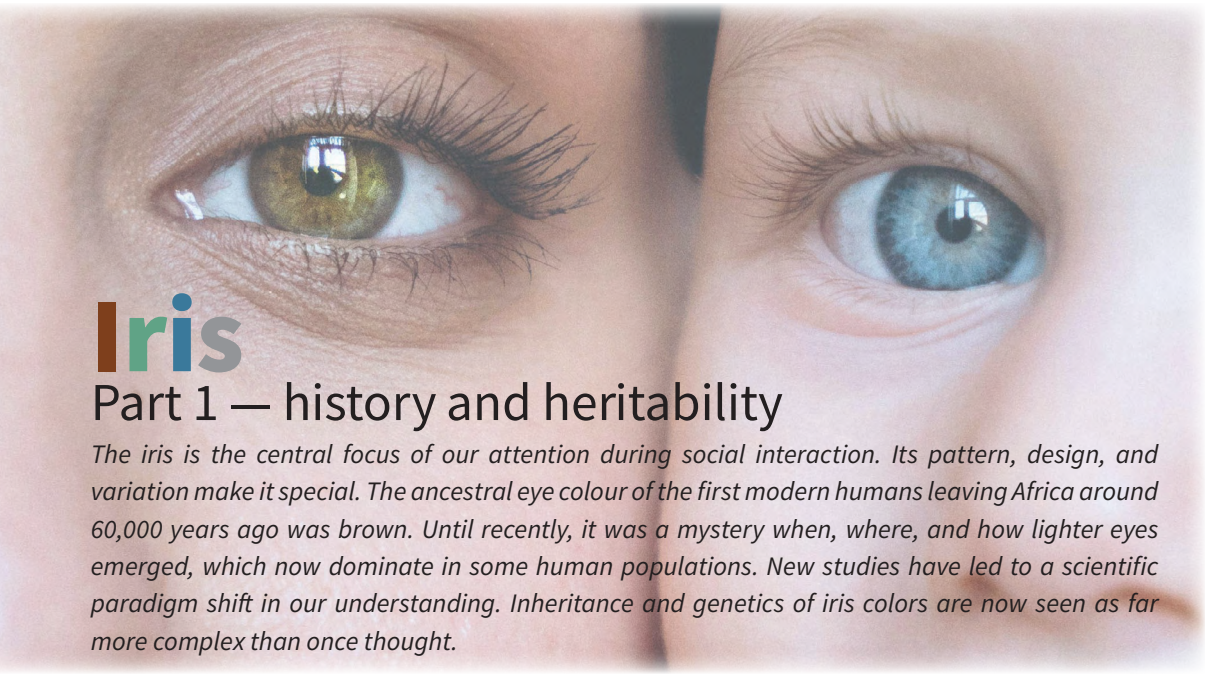

Iris
Part 1 — history and heritability

The iris is the central focus of our attention during social interaction. Its pattern, design, and variation make it special. The ancestral eye colour of the first modern humans leaving Africa around 60,000 years ago was brown. Until recently, it was a mystery when, where, and how lighter eyes emerged, which now dominate in some human populations. New studies have led to a scientific paradigm shift in our understanding. Inheritance and genetics of iris colors are now seen as far more complex than once thought.

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The dark-skinned Danish girl from the Stone Age

In 2015, an old piece of birch pitch with human bite marks was found on the Danish island of Lolland. Recently, the ability to obtain and reconstruct ancient genomes has improved dramatically. For this work, Swedish Svante Pääbo won the Nobel Prize in Medicine in 2022. Thanks to these advances, scientists could now extract genetic material from the lump and determine phenotypical features. The DNA came from a girl or woman—nicknamed "Lola" by the researchers—who lived about 5,700 years ago during the late Stone Age. Scientists then noticed something remarkable. She had blue eyes and dark skin and hair.[1] What was she doing in Denmark at that time?

We now know the answer. She was local with deep genetic roots in the area. New research has revealed that her kin, with the unique combination of blue eyes and a brown complexion, lived in vast parts of Europe. These so-called western hunter-gatherers (WHG) repopulated Europe around 11,000 to 12,000 years ago, following the Ice Age (**Figure 1**).

The new Europeans

The story becomes more complex in Northern Scandinavia, where migrating WHG from the south met fairer, but brown-eyed eastern hunter-gatherers (EHG), who moved in from the northeast after the ice melted. Notably, DNA from 18,000-year-old skeletal remains indicates that blondness originated separately in faraway Siberia. Consequently, 8000-year-old individuals with mixed ancestry have been identified in Motala, Sweden, who, for the first time, had probably both blue eyes and light hair.[2]

During Lola's lifetime, new Neolithic immigrants from Turkey had started to settle down in Europe and Scandinavia, with light skin and brown eyes. They introduced agriculture and rapidly replaced most of the WHG. Shortly after these farmers arrived, yet another momentous change shaped the demographics of Eurasia. From about 4,800 years before present (at the same time as the first Egyptian dynasties flourished), these early farmers were ousted in Denmark and, to a varying extent elsewhere, by a large wave of nomadic herders. Violence and perhaps the spread of infectious diseases played significant roles. A substantial proportion of the invaders had blonde hair and blue eyes, due to their shared EHG and WHG ancestry. Following this final and dramatic population turnover, the Danes, for example, have not changed substantially thereafter. The newcomers were from the Pontic-Caspian steppes of Ukraine and Southern Russia,

Figure 1. Meet Lola from Denmark. Modern Europeans carry genes for blue eyes from the indigenous, brown-skinned western hunter-gatherers, but only a smaller proportion of the total present DNA is inherited from them. Painting by Atle Einar Østern

where they domesticated horses and invented wagons. We call them the Yamnaya people.[3] They almost certainly introduced the Indo-European languages, which now dominate from Ireland to India in the Old World, **Box 1**. The only deviating languages are now found in the Basque region of Spain and Southern France, as well as in the areas with Finnish-Ugrian speakers today (which include the Samis, Finns, and Hungarians).

Hence, without going into the polarized debate about immigration, the new startling discovery is that what many associate with a typical pale European look (and often "ol' blue eyes") is the result of precisely that, relatively recently.

The first blue eyes

The question remains whether the WHG were truly the first people to have lighter eyes. Some studies suggest otherwise, estimating that blue eyes originated as a mutation around 42,000 years ago in the Near East.[4] If this is correct, everyone with this eye color might share a common ancestor. Descendants later migrated to the Caucasus and South-East Europe, where individuals had blue eyes at least 13,000–14,000 years ago.[5] From these lines, the WHG emerged.

Prior to the appearance of blue eyes, all humans had dark irises. In Africa, the continent of human origin, brown eyes protect the retina against the harmful effects of UV radiation. Moreover, brown eyed individuals may have a faster reaction time, which can be beneficial for activities where speed matters. In contrast, the disadvantage of depigmented irises is glare, because they allow 100 times more light to reach the retina.

Box 1. The word for eye is derived from Proto-Indo-European *h3ókws in related languages:

Ancestral language		*Modern versions*
Proto-Germanic:	augô **	Auge (German), auga (Icelandic), øye/øje/öga (Scandinavian)
Latin:	oculus	ojo (Spanish), occhio (Italian), œil (French)
Ancient Greek:	óps, osse	optical, ophthalmology
Proto-Slavic:	ǒko	oko (Polish)
Old-Armenian:	akn	ach´k´ (Armenian)
Proto-Albanian:	asī	sy (Albanian)
Proto-Indo-Iranian:	Hákši	akk^{h} (Punjabi), ānkh (Urdu), æsa (Sinhalese)
Proto-Celtic:	enek^{w}o (in+eye) = face	

*** Finnish silmä is unrelated, but aukko (hole) is possibly borrowed from Proto-Germanic*

Here you can listen to how the pronunciation of the word "eye" has changed over 6000 years:

- https://www.youtube.com/shorts/rWxWj6FTHvk
- https://www.youtube.com/watch?v=3eGRNu3Tzso

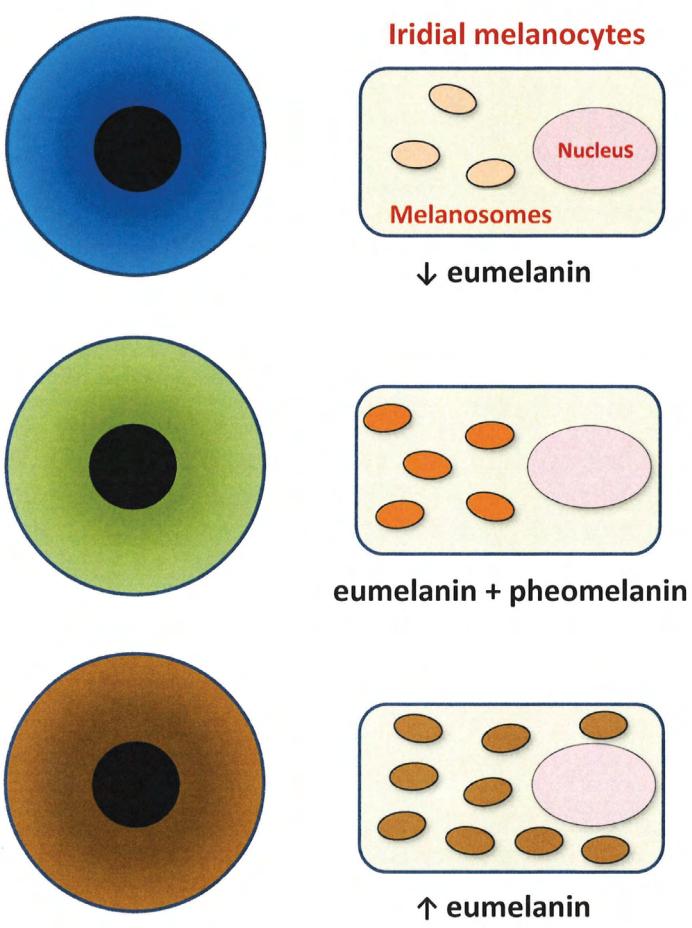

Figure 2. In all eyes, the number of melanocytes in the anterior border layer and iris stroma is similar. It is the quantity of intracellular melanosomes and their content of eumelanin (dark pigment) versus pheomelanin (yellow-red pigment) that determines the coloration of the iris. Illustration: Atle Einar Østern

Given this background, as brown eyes are in some ways superior, why did the bluish coloration then become so common in some parts of the world? Genetic drift (random change over time) and founder effect (establishment of a new population by a few individuals with deviant traits) are possible explanations. Beyond this, one hypothesis is that blue eyes could have potentially alleviated seasonal depression in dim conditions at higher latitudes. The problem with this explanation is, among others, that blue irises appeared before dark-skinned humans migrated northward and into cold Europe.

Could, then, a light iris rather be an asset in daily life? It has been demonstrated that blue-eyed people may perform better than brown-eyed individuals in muted light. Thus, better visual acuity in low-light situations, because more stray light enters the eye, could have made it easier to distinguish shapes. However, the question remains whether that is sufficient to offset the adverse effects compared to retaining sun-shielding brown eyes.

Therefore, another hypothesis has been proposed that the frequency of blue eyes spread fast in an ancestral population because carriers of this genotype and phenotype were more often chosen as mates. This is in accordance with "Fisherian runaway," which refers to a self-reinforcing process when a particular biological "ornament" is preferred and strongly selected, like, for instance, the oversized and otherwise non-adaptive peacock tail.[6]

The intricate genetics and distribution of eye colors

This brings us to genetics. The primary gene that determines eye color is the *OCA2* polymorphism at the chromosomal band 15q13.1. It produces a P-protein that

takes part in the production of the pigment. Malfunction here leads to melanin absence and oculocutaneous albinism, where the red blood vessels in the iris show through.

No blue or green eye pigments exist. Iris color depends on the amount and type of melanin. Blue eyes have the least melanin, brown the most, and green and hazel are in between (**Figure 2**). The diversity is immense. An uneven distribution of melanin can also occur naturally within the same iris. Different shades are also determined by the thickness and density of the iris, the extent of accumulated white collagen fibers, and tissue loss. In the end, several classification systems have been proposed to categorize the various colors found in human irises.

An iris appears blue for the same reason as the sky. Light with longer wavelengths passes through, while the shorter ones are reflected and scattered by the opaque layers. This is called the Tyndall effect.

Scientists previously thought that eye color inheritance followed Mendelian genetics. This meant blue was recessive and brown was dominant. Average outcomes could then be calculated and shown in pedigrees. Or so they believed.

But the truth is much more complicated. Newer research shows that many genetic loci take part in the multi-step process of melanin production and adjustment.[7] *HERC2* is crucial because it controls the nearby OCA2 gene through chromatin remodeling. Other genes, like *TYR, ASIP, SLC45A2,* and *SLC24A4*, contribute as well. Although *OCA2* accounts for 74% of eye color variation, all these genes may boost or reduce the level of melanin. Moreover, environmental factors may also influence melanin production. Contrary to the widely held belief, blue-eyed couples can have children with brown eyes.

Eye color can also change physiologically during a lifetime. The melanocytes need light under the influence of sympathetic innervation to start production of melanin following birth. In a study, 27% of newborn babies with blue eyes turned naturally brown at the age of 2 years, 7.5% became hazel, and 5% green.[8] This observation aligns with the phenomenon that numerous mammals, including kittens, are born exhibiting temporary shades of blue. (Birds, on the other hand, display extensive alterations and a range of other iris colors, like red and yellow, due to pigments such as carotenoids, purines, and pteridines).

The frequency of distinct eye colors varies in different countries (**Box 2**). Worldwide, 70%–80% of humans have brown eyes, 8%–10% blue, 5% hazel, 5% amber, 3% gray, and 2% green eyes. Furthermore, men are much more prone to have blue eyes, possibly representing a gender-related factor.[9]

Box 2. Percentage of eye colors in selected countries:

Country	Brown eyes	Blue eyes	Intermediate eyes
Iceland	9%	75%	14%
Denmark	15%	65%	21%
Netherlands	22%	61%	11%
Germany	27%	40%	33%
United Kingdom	32%	43%	26%
France	34%	22%	44%
Spain	75%	11%	13%
Uzbekistan	91%	3%	6%

Source: https://worldpopulationreview.com/country-rankings/eye-color-percentage-by-country

Legacy of the past

If you immerse yourself in someone's gaze, or your own reflection in the mirror, the iris reveals itself as something unique: vividly magical and unlike anything else. Behind these sparkling anatomical structures lies a fascinating story about a long and winding journey that science is only now beginning to fully uncover. Along the way, there have been surprising and dramatic twists. Ultimately, the appearance of one of the most minor yet most captivating parts of the human body has been shaped over time by a complex interplay of ancient migrations, social preferences, environmental pressures, and the unpredictable randomness of genetic change. The result is a mosaic of hues that remains visible at every glance.

***This series on the Iris will be continued in coming issues of* Oftalmolog.**
Up next:

- The importance of the iris to human well-being

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