Consanguinity & Eye diseases

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Consanguinity or kinship ("blood relation", from the Latin word *consanguinitas*) is characterized by the sharing of common ancestors. Consanguineous persons have at least one common ancestor in the preceding few generations. A consanguineous marriage is one in which two individuals related by blood, such as cousins, get married and have children.
Around the globe, consanguineous marriages have been practiced by many societies from time immemorial. It is widely practiced in Asia, North Africa, Switzerland, Middle East, some parts of China, Japan and fishermen communities in Europe and America. One in two rural marriages in Tamil Nadu and Andhra Pradesh are consanguineous.

Genes, which occur in pairs, are packages of information that we inherit from our parents. The risk of birth defects is higher in consanguineous marriages because there is a greater chance of two related individuals sharing a common harmful gene and both passing it on to the child (Autosomal recessive inheritance). The risk and type of birth defects in consanguineous marriages vary depending on how closely the couple is related and also based on defective gene expression. Consanguineous couples can also have normal healthy children.

Consanguineous marriage occurs between first cousins, uncle's son marries auntie's daughter or vice versa and Maternal uncle marries his niece (sister's daughter). Among these the risk of having a genetically defect child is higher in the latter type of marriage than the former.

Consanguinity leads to:

- ✓ Death of infants before, during or immediately after birth, increased incidence of birth defects. The most common defects are in the sense organs and the nervous system.
- ✓ Blood cancer (acute lymphocytic leukemia).
- ✓ Breathing problems for children at birth (apnea).
- ✓ Increased susceptibility to disease.

Consanguinity in eye diseases:

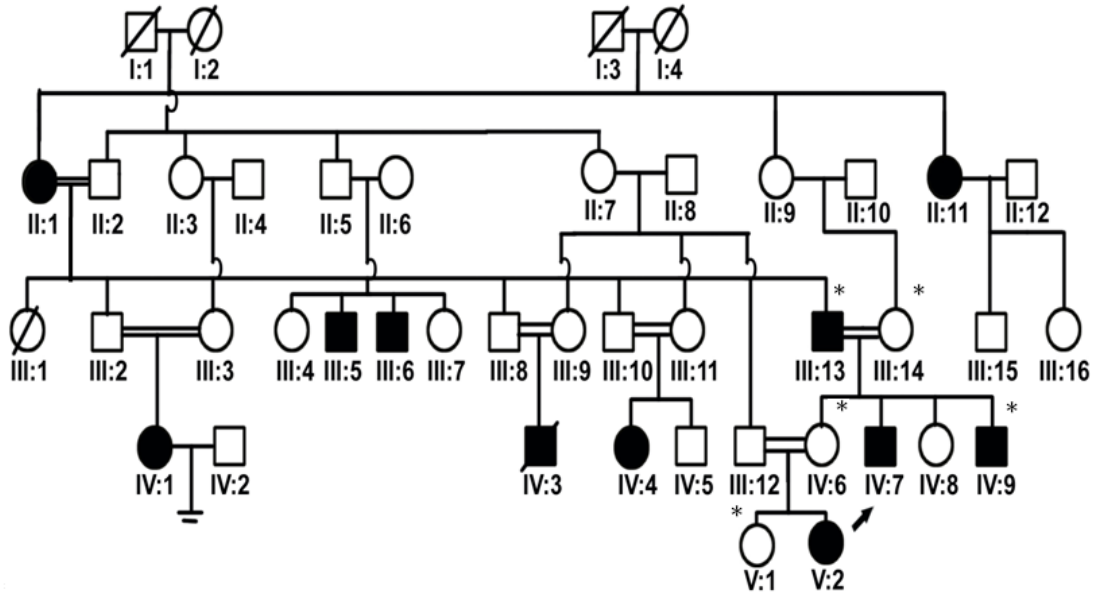
- ✓ Squint in children.
- ✓ A congenital condition (present at birth) where the iris is absent - **Aniridia.**
- ✓ Atrophy (Inefficiency) of the retina, Night blindness - Retinitis **Pigmentosa.**
- ✓ Pigmentation of the hair, skin and eyes - **Oculocutaneous Albinism.**
- ✓ The eyes without pigmentation - Ocular **Albinism.**
- ✓ Photophobia (aversion to light), decreased visual acuity, nystagmus (constant jerky movement of the eyes)
- ✓ Due to developmental abnormalities absence of either Iris/choroid structures in the eye – **Coloboma.**
- ✓ Severely impaired vision - **Leber congenital amaurosis.**
- ✓ Vision loss due to optic nerve damage - **Glaucoma.**
- ✓ Along with retinitis Pigmentosa, obesity, mental retardation, kidney failure etc - **Lawrence-Moon-Bardet-Biedl syndrome.**
- ✓ Macular degeneration (damage to the retina) - **Stargardt disease.**
- ✓ Deaf blindness - **Usher syndrome.**

Why do consanguineous marriages result in hereditary diseases?

Relationship	Average % of DNA shared
parents to children	50%
brothers and sisters	50%
uncle and niece	25%
first cousins	12.5%

Due to the sharing of genetic material from the common ancestor, the marriages between uncle, niece and first cousins results in genetic disorders in the offspring. Hence creating awareness for avoiding consanguineous marriages is the foremost preventing measures.

A typical family chart representing the consanguineous marriage of Ocular Albinism:



Unaffected male

Affected male

Unaffected female

Affected female

Deceased

Due to the consanguineous marriages in this family between the individuals II:1 & II:2 , III:2 & III:3, III:8 & III:9, III: 10 & III:11, III: 13 & III: 14, III:12 & IV:6 most of their children were affected with Ocular albinism.