Link between Consanguinity and Genetic Diseases

Why in news?

Many key insights that are biomedically relevant have been unearthed by looking through the lens of consanguinity.

What is consanguinity?

- **Consanguinity** – It is the practice of marrying close relatives, an age-old tradition that is still practised widely in several human societies.
- It is the kinship characterized by the sharing of *common ancestors.*

Kin are of two basic kinds: consanguineous (sharing common ancestors) and affinal (related by marriage).

- It is a social as well as genetic construct.
  - **Social context**- Marriage between individuals related by blood.
  - **Genetic context (inbreeding)-** Marriage between genetically related individuals.
- Studies have found that a significant fraction of the global population practises consanguinity and that that has increased the mortality and the rate of recessive genetic diseases in these peoples.
- **Global status** - There is an evidence that suggest those of the Egyptians and Incas, among others, could have practised inbreeding or consanguinity.
- **Status of India-** With more than 4,000 endogamous (groups people marrying within the same caste/tribe or group), India has been a fertile ground for consanguinity.

According to one estimate, approximately 15-20% of the world's population practises inbreeding, especially in Asia and West Africa.

What are the benefits of consanguinity?

- **Social impact**- It leads to
  - Stable marital relationships
  - Improved female autonomy
  - Less domestic violence and lower divorce rates
- **Economic impact**- It
  - Reduces the instances of dowry and the preservation of landholdings
  - Reduces the risk of family financial problems
- **Cultural impact-** Consanguinity provides opportunities for the transmission of

cultural values and cultural continuity
    - Better compatibility with in-laws
    - Ease of marriage arrangements
- **Genetic traits-** Consanguinity may cause homozygosity of alleles in offsprings, preserving the advantageous genetic traits in human as well as animal populations.
- **Disease protection**- Protection against disease has been postulated because of the selection of protective genotypes or homozygosity of protective alleles.
- Parental consanguinity seems to protect against breast cancer in the Tunisian population.
- The protective role of consanguinity against multiple sclerosis has also been reported.
- **Health-** Consanguineous families are helpful in the identification of novel disease gene by linkage analysis.

What is the link between consanguinity and diseases?

- **Recessive disease**- Many modern consanguineous societies, like the Amish population in the U.S., have been studied for recessive diseases.
- **Autozygosity-** Scientist have extensively used autozygosity as an approach to identify new genetic diseases in populations where consanguineous marriage practices is the norm.
- **Disease-** The results of these studies have helped us uncover previously unknown genetic diseases as well as estimate different population's genetic predisposition to common diseases.
    - Recent study suggested that consanguinity could increase the risk and the rate of diseases like type-2 diabetes.
- **Genomic research-** Advances in genomic research will provide innovative solutions to mitigate the risks associated with consanguinity or genetic diseases.
- **Genetic diagnostics-** The genomic research would result in personalised medicine, genetic diagnostics and genetic counselling that will play a pivotal role in improving the health outcomes of affected individuals and their families.

What lies ahead?

- Improved family finances, public health awareness, and higher female education can decrease consanguinity rates.
- Premarital genetic screening should be offered for the most common population-specific genetic disorders.
- Physicians should be educated on the importance of focusing on consanguinity-associated diseases like hearing loss, thalassemia, cystic fibrosis, intellectual disability, etc.

References

1. The Hindu- Unravelling links between consanguinity and genetic diseases
2. NIH-National Human Genome Research Institute



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