Abstract Title: Consanguinity: a cause for burden of rare genetic disorders

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Abstract: Consanguinity, which refers to the marriage or reproduction between close blood relatives, can indeed be a cause for the increased burden of rare genetic disorders. When close relatives have children, there is a higher likelihood that both parents carry the same recessive genetic mutations, increasing the risk of these mutations being expressed in their offspring. This can lead to a higher prevalence of rare genetic disorders within such populations. However, it's important to note that the impact of consanguinity on genetic disorders can vary depending on the specific genetic makeup and cultural practices of different populations.

Consanguinity, or marriage between close blood relatives, is practiced to varying degrees in India. It's more common in certain regions and communities than in others. This practice is influenced by cultural, social, and historical factors. In some parts of India, particularly in rural areas, it's more prevalent due to factors such as family traditions, limited social interactions, and arranged marriages within the same caste or community.

Consanguineous marriages in India can lead to a higher risk of offspring inheriting rare genetic disorders due to the increased likelihood of both parents carrying the same recessive genetic mutations. However, it's important to emphasize that not all consanguineous unions result in genetic disorders, and the risk varies depending on the genetic background of the individuals involved.

Efforts to raise awareness about the potential health risks associated with consanguinity and to promote genetic counseling have been made to address these concerns and reduce the burden of genetic disorders in such populations.

Several genetic disorders are prevalent in India, including:

1. Thalassemia

2. Sickle Cell Anemia

3. Cystic Fibrosis

4. Hemophilia.

6. Phenylketonuria

7. Muscular Dystrophy

8. Wilson's Disease

The prevalence of these disorders can vary by region and population group in India. Efforts are being made to increase awareness, diagnosis, and management of these conditions to reduce their impact on affected individuals and families.

The coefficient of consanguinity is a measure that quantifies the degree of relatedness or consanguinity between two individuals. It is used in genetics to estimate the probability that two alleles at a specific gene locus are identical by descent due to a common ancestor.

The coefficient of consanguinity can be calculated using a formula that takes into account the specific relationship between the two individuals in question. The formula varies depending on the degree of consanguinity, such as first cousins, second cousins, and so on.

For example, in a first cousin marriage, the coefficient of consanguinity is 1/8, indicating that there is a 1/8 chance that a specific gene locus is identical by descent between the cousins. In a second cousin marriage, the coefficient of consanguinity is 1/32, and so on.

This coefficient is important in understanding the genetic risks associated with consanguineous marriages, especially when it comes to the increased likelihood of offspring inheriting rare recessive genetic disorders due to shared ancestry.

Consanguineous marriages, or marriages between close blood relatives, are a complex topic with both advantages and disadvantages. It's important to note that the advantages and disadvantages can vary depending on the specific cultural, social, and genetic context. Some potential advantages of consanguineous marriages can include:

1. Cultural and Social Factors

2. Social and Economic Ties

3. Compatibility

4. Reduced Dowry Pressure:

5. Kinship Support

It's important to consider that while there are potential advantages, there are also significant health risks associated with consanguineous marriages, particularly in terms of an increased risk of offspring inheriting rare genetic disorders due to the sharing of recessive genetic mutations. The potential advantages and disadvantages should be weighed carefully, and genetic counseling is often recommended to assess and mitigate these risks.

Analysis of our small data at our hospital supports this cause

Area of expertise: Maternal fetal medicine, prenatal diagnosis, fetal interventional procedures, perinatal genetics, high ris preg.