CONSANGUINITY AND HEALTH

Hatem El-Shanti
CONSANGUINITY AND HEALTH

DEFINITION

- Marriage between blood relatives
- Marriage between individuals who are second cousins or more closely related
- More distantly related marriage can still be significant especially in highly inbred families
Coefficient of kinship ($\Phi$) of two individuals is the probability that a gene chosen at random in the first individual is identical by descent to the gene at the same locus in the second individual.
Coefficient of inbreeding (F) of an individual is the coefficient of kinship of his/her parents; in other words: It is the probability that the two genes which the individual has at a given locus are identical by descent.
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FIRST COUSIN MARRIAGE

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SECOND COUSIN MARRIAGE

I

II

III

IV

V

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EXTENSIVE INBREEDING

CMA

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CONSANGUINITY RATES

- 20 % to 70 %
- Not a religious practice
- First cousin marriage for Moslems
- Second cousin marriage for Christians
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PROMOTING FACTORS

- Tradition
- Economic reasons
- Psychosocial and family support
- Geographical
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EFFECTS

- Autosomal recessive disorders
- Birth defects
- Perinatal mortality
- Fertility
- Fetal wastage
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GENE MAPPING & IDENTIFICATION

- Progressive pseudorheumatoid dysplasia
- Wolfram syndrome type II
- Majeed syndrome
- Acrodermatitis enteropathica
- Selective vitamin B12 deficiency
- Murine chronic multifocal osteomyelitis
- Progressive myoclonic epilepsy – ataxia syndrome
- Isolated ectopia lentis
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MYOCLONIC EPILEPSY – ATAXIA

I

II

III

IV

V

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MAPPING TO CHROMOSOME 12
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MUTATION

![Graph showing normal, carrier, and mutant states with a mutation indicated by an asterisk. The graph compares DNA sequences with peaks representing different nucleotides. The normal state does not show a mutation, the carrier state has a single mutation, and the mutant state shows multiple mutations.](image)
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PREVENTIVE STRATEGIES

- Awareness and public education
- Carrier testing
- Premarital and preconception testing
- Genetic counseling
- Prenatal diagnosis
- Pre-implantation diagnosis
To err is human.
But to completely mess up you need a computer.