ISSN 0976-5417 Cross. Res. June 2014 Vol. 5 No.1

Parental Consanguinity and Genetic disorders with reference to Asaripallam, India.

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Abstract

Consanguineous marriage is common in many societies, which leads to an increased prevalence of recessive disorders. The aim of the present study was to determine the prevalence of genetic disorders and patterns of consanguinity in an inbreeding human population. A sample of 111 families of a single endogamous population of Asaripallam was studied based on information on marital pattern and diseases. The marriage celebrated in all families was between relatives. First cousin marriages were encountered in 11%, second cousin in 55%, distant relation in 34% of the families respectively and many families showed the presence of disorders. The disorders observed are diabetes, cerebral palsy with mental retardation, arthritis, vision problems and blood complications. This study emphasizes the role of homozygous recessive gene in the causation of genetic disorders.

Key words: Inbreeding, consanguineous marriages, genetic disorders

Introduction

Inbreeding is defined as the mating between closely related individuals. The study and consequences of inbreeding are considerable concerns in the field of genetics. A marriage is said to be consanguineous, where the marriages are solemnized among persons descending from the same stock with close biological relations. Consanguineous marriages have been practiced for hundreds of years in many parts of the world. Marriage between two such individuals who have at least one traceable common ancestor is said to be 'consanguineous' and offspring of such mating 'inbred'. Traditionally, some cultures have practiced and continue to practice marriage between relatives as a means of strengthening family ties and retaining property within the family. Due to consanguineous marriages there are many genetical defects in the offspring. When a person marries within the community to a person who may also have such a family defect, the child inherits two copies of this faulty gene, and thus has the defect. But when a person marries outside the community, a person brings in genes from a much larger gene pool, and the odds that the child will inherit the problem reduce remarkably.

Worldwide, nearly 1,000 million people live in countries where 20 to more than 50% of marriages are consanguineous. The existences of consanguineous marriage and genetics perspective on consanguineous marriage in different population of the world have been studied by many authors [1-10]. Although a high proportion of marriages in Kanyakumari are consanguineous (i.e. contracted between close biological relatives), with few exceptions, there is a little knowledge on the association between consanguinity and genetic diseases. So an attempt was carried out to overview the prevalence of consanguineous marriages in Asaripallam, a village known for the same where marriage outside the community leads to excommunication to analyze the genetic diseases of this inbreeding human population.

Methods

Asaripallam a village of Kanyakumari District, India noted for consanguineous marriages was selected for this study. Primary data on name, age, relationship of the couple, name and age of their children, disorders from the family members were collected by face-to-face interview.

The possible types of marriage pattern, i.e., matrilateral and patrilateral cross-cousin marriage, relationship between the spouses and the percentage of the different types of consanguineous marriages of the population were assessed.

Results

The study covers a total of 111 families of Anuripallam (Table - 1). They showed different patterns of consanguineous marriages. First cousin marriages were encountered in 11%, second cousin in 55%, distant relation in 34% of the families. Second cousin marriages (55%) were the most common form of consanguineous union favoured (Table - 2). Many families showed the presence of disorders (Table - 3). At least one family member of each family showed the presence of diabetes.

Table - 1: Age wise analysis of males and females in the study population

Age group (year)	Total	Gender	Number	Percentage	Total percentage
0-10	48	Male	23	7.28	15.19
		Female	25	7.91	
10-20	27	Male	14	4.43	8.54
		Female	13	4.11	
20-30	69	Male	29	9.18	21.84
		Female	40	12.66	
30-40	66	Male	36	11.39	20.89
		Female	30	9.49	
40-50	54	Male	33	10.44	17.09
		Female	21	6.65	
50-60	25	Male	10	3.16	7.91
		Female	15	4.75	
60-70	19	Male	13	4.11	6.01
		Female	6	1.89	
70-80	7	Male	4	1.27	2.22
		Female	3	0.95	
80-90	1	Male	1	0.32	0.32
		Female	0	0	
Population size	**316**	**Male**	**163**	**51.58**	**100**
		Female	**153**	**48.42**	

Table - 2: Parental Consanguinity in the study population

Relationship	Number	Percentage
First cousin	12	11
Second cousin	61	55
Distant relation	38	34

Table - 3: Genetic disorders observed in the study population

Disorder	Number	Percentage
Arthritis	7	6.3
Blindness	3	2.7
Cerebral Palsy with mental retardation	3	2.7
Handicap	2	1.8
No Children due to Blood complication	1	0.9

Discussion

Inbreeding is reproduction from the mating of pairs who are genetically closely related. Inbreeding results in homozygosity which can increase the chances of offspring being affected by recessive or deleterious traits [6]. This generally leads to a decreased fitness of a population [11] which is called inbreeding depression. The avoidance of expression of deleterious recessive alleles caused by inbreeding is thought to be the main selective force maintaining the out crossing aspect of sexual reproduction [12]. Inbreeding may result in a far higher phenotypic expression of deleterious recessive genes within a population than would normally be expected [13]. As a result, first-generation inbred individuals are more likely to show physical and health defects.

Inbreeding can occur just because a small population has been isolated during some time [13 & 14]. It can also occur in a large population if individuals tend to mate their relatives as a method of forming political alliances among elites, to protect property, wealth and position. In societies where close relatives marry, these unions tend to provide avenues for families to retain wealth and land within bloodlines. For hundreds of years, inbreeding was historically unavoidable in Iceland due to its then tiny and isolated population [15]. Marriage customs are the important components of social structures [16]. Attempts to break up marriage customs with long traditions should not be initiated unless all possible social consequences have been carefully considered. In this study we observed that matrilateral cross-cousin marriage is more prevalent type of unilateral cross-cousin marriage [17].

Autosomal recessive disorder occurs in individuals who have two copies of the gene for a particular recessive genetic mutation [18]. It is more likely that related parents will both be carriers of the same recessive gene, and therefore their children are at a higher risk of a genetic disorder. The extent to which the risk increases depends on the degree of genetic relationship between the parents. The risk is greatest when the parents are close relatives and lower for relationships between more distant relatives, such as second cousins, though still greater than for the general population [19]. Children of parent-child or sibling-sibling unions are at increased risk compared to cousin-cousin unions. This is due to the presence of recessive disease genes [2].

An Autosomal Recessive form of Spastic Cerebral Palsy (CP) with microcephaly and mental retardation was observed in two families in the study area with phenotypic variability among the affected individuals. Diabetes which is a recessive trait is another hereditary disease observed in the study area. At least one family member of each family showed the presence of diabetes. Patients from consanguineous matings with autosomal recessive nephrogenic diabetes insipidus homozygous for mutations in the aquaporin 2 water-channel gene were described by Van Liebrug *et al.* [20]. Arthritis, another common disease observed is caused by a combination of several factors including genetic makeup. In one family, the new born baby dead due to blood complication. A type of blood complication, known as spherocytosis, a collection of inherited disorders which manifest as spherical-shaped erythrocytes (spherocytes) on the peripheral blood smear may be the reason for the death of the baby.

Consanguineous marriage exists throughout human societies of the world. Inbreeding or consanguinity among human beings has both positive effects in terms of social and economic advantages for the couples and their families and detrimental effects on physical and mental health of the progeny. Effects of consanguineous marriage on human progeny related to child health, reproductive behaviour- still birth, pregnancy outcomes, its measurement, mortality and morbidity, and its effects on the mental health should be studied thoroughly. Indirect counseling may be a good prescription to avoid consanguinity.

Acknowledgements

The authors would like to express their sincere thanks to Mr. Jesu Raj, Department of Zoology, Holy Cross College, Nagercoil, for his kind support during data collection.

References

1. Bowirrat Abdalla and Armaly Zaher (2013). Consanguineous Marriages in the Middle East: Nature Versus Nurture, The Open Complementary Medicine Journal, 5: 1-101.
2. Bittles, AH. (2008). A community genetics perspective on consanguineous marriage. Community Genet., 11 (6): 324-330.
3. Riegler, A., Marroni, F., Pattaro, C., Gueresi, P. and Pramstaller, P. (2008). Isolation and marriage patterns in four South Tyrolean villages (Italy) during the nineteenth century. J. Biol. Sci., 40: 787-791.
4. Blanco Villegas, MJ. and Fuster, V. (2006). Reproductive pattern in consanguineous and non consanguineous marriages in La Cabrera, Spain. Ann. Hum. Biol., 33 (3): 330-341.
5. Hussain, R. and Bittles, AH. (2004). Assessment of association between consanguinity and fertility in Asian populations. J. Health Popul. Nutr., 22 (1): 1-12.
6. Nabulsi, MM., Tamim, H., Sabbagh, M., Obeid, MY., Yunis, KA. and Bitar, FF. (2003). Parental consanguinity and congenital heart malformations in a developing country. Am. J. Med. Genet., 116A (4): 342-347.
7. Fuster, V. (2003). Inbreeding pattern and reproductive success in a rural community from Galicia (Spain). J. Biosoc. Sci., 35 (1): 83-93.
8. Jaber, L., Halpern, GJ. and Shohat, M. (1998). The impact of Consanguinity worldwide, Commun. Genet., 1: 12-17.
9. Bittles, AH., Grant, JC. and Shami, SA. (1993). "Consanguinity as a determinant of reproductive behaviour and mortality in Pakistan". Int. J. Epidemiol., 22 (3): 463-467
10. Pattener, D. (1981). Inbreeding secular changes in the Riomaggiore and Silla valleys, Italy, from 1565-1980, Acta Anthropogenetica, 5: 271-278.

11. Jimenez, JA., Hughes, KA., Alaks, G., Graham, L. and Lacy, RC. (1994). An experimental study of inbreeding depression in a natural habitat. Sci., 266 (5183): 271-273.
12. Bernstein, H., Byerly, HC., Hopf, FA. and Michod, RE. (1985). "Genetic damage, mutation, and the evolution of sex". Sci., 229 (4719): 1277-1278.
13. Griffiths, Anthony, JF. Jeffrey H. Miller, David T. Suzuki, Richard C. Lewontin and William M. Gelbart (1999). An introduction to Genetic Analysis, New York: W. H. Freeman, 726–727.
14. Bittles, AH. and Black, ML. (2010). Consanguineous Marriage and Human Evolution. Annual Review of Anthropology, 39: 193-207.
15. Helgason, A., Palsson, S., Guthbjartsson, D., Kristjansson, T. and Stefansson, K. (2008). An Association Between the Kinship and Fertility of Human couples. Sci., 319 (5864): 813-816.
16. Vogel and Motulsky (1997). Human Genetics: Problems and approaches, 3rd edition, Springer, New York.
17. Levi-Strauss, C. (1969). The elementary structures of kinship. Boston: Beacon Press.
18. Hartl, DL. and Jones, EW. (2000). Genetics: Analysis of Genes and Genomes. Fifth Edition. Jones and Bartlett Publishers Inc., 105-106.
19. Kingston, HM. (2002). ABC of Clinical Genetics (3rd ed.), London, BMJ Books, 7.
20. Van Liebrug, AF., Verdijk, MA., Knoers, VV., Van Essen, AJ., Proesmans, W., Mallmann, R., Monnens, LA., Van Oost, BA., Van Os, CH. and Deen, PM. (1994). Patients with autosomal nephrogenic diabetes insipidus homozygous for mutations in the aquaporin 2 water-channel gene. Am. J. Hum. Genet., 55 (4): 648-652.