

CONSANGUINITY AND CHROMOSOMAL ABNORMALITIES

Lakshmi Prabha Subhash R ^{*1}, Anupama D ², Jayarama S Kadandale ³, Meenakshi Bhat ⁴, Harshal K L ⁵, Khizer Hussain Afroze M ⁶.

^{*1} Professor and HOD, Dept of Anatomy, Sri Siddhartha Medical College, Tumakuru, Karnataka, India.

² Associate Professor, Dept of Anatomy, Sri Siddhartha Medical College, Tumakuru, Karnataka, India.

³ Consultant cytogenetist, Division of cytogenetic, Dept of Anatomy, Sri Siddhartha Medical College, Tumakuru, Karnataka, India.

⁴ Genetic Counsellor, Division of cytogenetic, Dept of Anatomy, Sri Siddhartha Medical College, Tumakuru, Karnataka, India.

⁵ Research Associate, Division of cytogenetic, Dept of Anatomy, Sri Siddhartha Medical College, Tumakuru, Karnataka, India.

⁶ Research Scholar, Dept of Anatomy, Sri Siddhartha Medical College, Tumakuru, Karnataka, India.

ABSTRACT

Back ground: Consanguinity (CON) is defined as marriage between the close relatives and strongly favoured among the Populations of South India and plays an important role in the high incidence of congenital malformations in children, due to Expression of rare recessive genes inherited from a common ancestor.

Aims: The present study is undertaken to analyze the effects of CON on congenital malformation and associated chromosomal abnormalities

Methods and Material: A total of 550 cases with suspected genetic etiology were referred to Division of Cytogenetics, Department of Anatomy SSMC, Tumakuru, Karnataka since 2 years. Karyotyping was done from peripheral blood lymphocyte culture and G-T-G Banding using trypsin and Giemsa. Karyotype descriptions were reported and findings were statistically analyzed and those patients with chromosomal abnormalities received post Cytogenetic Genetic counselling in our Department.

Results: CON marriages were represented in 36% of cases. Stillbirths, recurrent abortion, and congenital anomalies were significantly increased. Chromosomal anomalies were grouped as structural and numerical anomalies and highest frequency of abnormal karyotype was found among cases of Down's syndrome and repeated abortion.

Conclusions: The present study is undertaken to analyze the effects of CON on Genetic disorders and associated chromosomal abnormalities which demonstrate the importance of cytogenetic evaluation, public health education, and genetic counseling.

Key-words: Consanguinity, Congenital anomalies, chromosomal anomalies, Nondysjunctions, Autosomal recessive.

Address for Correspondence: Dr. Lakshmi Prabha Subhash R, Professor and HOD, Dept of Anatomy, Sri Siddhartha Medical College, Tumakuru, Karnataka, India. **Mobile phone no:** +91- 9620769595
E-Mail: lakshmiprabha100@yahoo.com

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INTRODUCTION

Consanguinity refers to the quality of being descended from the same ancestor. Consanguinity was seen in 29.14% of cases and has been described as an important factor contributing to increased congenital malformations [1].

Congenital malformations (CM) began to emerge as one of the major childhood health problems and it refers to any abnormality, genetic or not which is present at birth. The etiology of congenital malformations is genetic in 30-40% and environmental in 5-10% of cases. In genetic disorders, chromosomal anomalies constitute 6% of cases [2].

Globally 20% of human population live in communities with a preference for consanguineous marriage and among the consanguineous families the blood relationship of uncle niece is 22.5%, seems to have highest risk of affecting the offspring [3]. Genetic effects of Consanguinity can be traced to the fact that the inbred individual may carry 2 copies of a gene that was present in a single copy in the common ancestor of consanguineous parents. A recessive gene may thus come to light for the first time in an inbred descendent after having remained hidden for generations, influencing the inheritance of genetic disorder [2]. Chromosomal analysis is an important component to the diagnosis and evaluation of congenital anomalies, developmental delay and intellectual disabilities and affects 7.5% of all conceptions [4].

This study is aimed at determining the role of Consanguinity on Congenital malformations and its association with chromosomal abnormalities.

MATERIALS AND METHODS

A total of 550 cases with suspected genetic etiology were referred to Division of Cytogenetic, Department of Anatomy SSMC, Tumakuru, Karnataka since 2 years. A detailed medical history and informed consent was obtained with all cases before cytogenetic analysis. Suspected genetic cases were Bad obstetric history (BOH), Mental Retardation (MR), multiple congenital anomalies, Down's syndrome, primary amenorrhea and infertility referred for Karyotyping and genetic counselling. H/O congenital malformation in other offspring and members of their Fam-

ily and parental consanguinity was noted. Data on consanguinity was traced from family pedigree up to a Minimum of three generations.

Karyotyping was done from peripheral blood lymphocyte culture and G-T-G Banding using trypsin and Giemsa. Minimum of 30 cells were routinely analysed from the best metaphases. Karyotype descriptions were reported and findings were statistically analyzed and those patients with chromosomal abnormalities received post Cytogenetic Genetic counselling in our Department.

RESULTS

Couples with BOH and Down's syndrome are the 2 main groups referred to the Lab. The next common referrals were intellectual disability, congenital anomalies and others. The highest frequency of abnormal Karyotype was found in Down's syndrome followed by BOH, congenital anomalies and intellectual disability.

Table 1: Showing cytogenetic study in study population.

	Total Referral Number	Abnormal Karyotype		Consanguinity
		Number	Percent (%)	
Down's	41	39	95.10%	13
BOH (Couples)	132	8	6.20%	54
Congenital anomalies	118	3	2.50%	34
Intellectual disability	82	2	2.40%	12
Female infertility	26	1	3.80%	9
Male infertility	12	0	0%	7

Out of 550 cases, 198 were consanguineous (36%) and 352 cases were non consanguineous (64%). The most common degree of consanguineous marriages among our patients was first cousins 51.7% and uncle niece 48.2%. Association of Consanguinity with chromosomal abnormalities is summarised in table 1

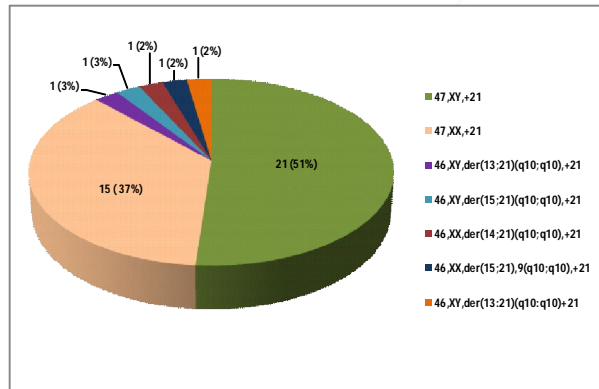
Table 2: Showing frequencies of different forms of abnormal karyotype.

Cases	Cases with consanguinity		Cases with non - consanguinity		Total Number
	Number	Percent (%)	Number	Percent (%)	
All	198	36%	352	64%	550
Normal karyotype	179	90.40%	317	90%	496
Abnormal karyotype	19	9.60%	35	9.90%	54
Numerical abnormality	13	68.40%	25	71.40%	38
Structural abnormality	6	31.50%	10	28.60%	16

Down's Syndrome: Among the 550 cases, 41 cases (7.45%) were Down's syndrome (25 males

and 16 females). The chromosomal abnormalities were free Trisomy seen in 33 cases (80.4 %), translocation in 5 cases (12.1%) and normal variant seen in 3 cases (7 %). Consanguinity was seen in 13 cases (31.7%), Out of which Numerical abnormality seen in 11 cases and structural abnormality seen in 2 cases.

Fig. 1: Showing chromosomal abnormalities in down's syndrome.



Bad Obstetric History: Of total 550 cases, BOH is seen in 132 couples. Mean maternal age at first conception was 22.2 years, ranging from 15- 36 years. First cousin marriage is seen in 54.16% and uncle niece marriage accounts for 37.5% and far relatives in 8.3% of cases.

Cytogenetic evaluation of these couples's revealed chromosomal variations in 8 cases (3%) of which 3 were males and 5 were females. The variations include 3 inversions (37.5 %), 2 translocations (25 %) and 3 heterochromatic variations (37.5%). The most frequently affected chromosome was chromosome 9. 54 couples had consanguineous marriage (40.9%) and 5 cases had structural anomalies associated with Consanguinity (4.6%). BOH with consanguinity and abnormal karyotype presented as pregnancy loss (abortions), stillbirths, IUDs and Congenital malformations.

Table 3: Showing chromosomal aberrations in couples with BOH.

Type of Abnormality	Number of cases	Number of pregnancy loss			Consanguineous		Non Consanguineous
		1 st trimester	2 nd trimester	Others	1 st cousin	Uncle niece	
45,XX,der(14;21)(q10;q10)	1				-	-	+
46,XX,t(3;4)(p13;q33)	1	3	-	-	-	+	-
46,XX,inv(6)(p22;q13)	1	-	-	2 IUD	+	-	-
46,XY,9qh+	1	-	2	1	-	+	-
46,XY,inv(9)(p11q,13)	1	2	-	-	-	-	+
46,XY,15p+	1	3	-	-	-	+	-
46,XX,inv(9)(p11,q13)	1	3	1	-	-	-	+
46,XY,9qh+ breakage	1	-	-	-	-	-	+

Congenital Anomalies: Congenital anomalies were seen in 118 cases (21.2%). Of these Consanguinity was noted in 34 cases (28.8%). Eye and renal anomalies were the most prevalently seen. Neural tube defects, skeletal dysplasia, polythelia, Meckler Gruber syndrome, cleft lip and cleft palate were other cases referred. Cleft lip and cleft palate and polydactyl showed chromosomal anomaly of 9qh+ and inversion 7 (Table 4). In our study 7 cases were referred as Ambiguous genitalia (5.9%), 5 showed 46, XY karyotype and one showed 46, XX karyotype. Two cases were associated with Consanguinity. Of 21 cases (3.8%) of Polydactyl were referred, 4 cases (19%) were associated with consanguinity. 1 case was associated with chromosomal anomaly as inversion 7.

Table 4: Showing prevalence of congenital anomalies.

Malformation system	Frequency	Percent (%)	Consanguinity
Eye anomalies	16	13.50%	7
Polydactyl	21	18%	3
Muscular skeletal system	6	5.10%	3
Neural tube defects	7	5.90%	2
Renal anomalies	8	6.80%	2
Ambiguous genitalia	7	6%	2
Facial anomalies	5	4.30%	2
CTEV	4	3.40%	2
Cardiovascular system	4	3.40%	1
Undescended testis	3	2.60%	1
Vaginal atresia and uterine anomalies	2	1.70%	1
Cleft lip and palate.	5	4.30%	1
Gastroschisis	2	1.70%	-
Polythelia	1	0.85%	-
Orofacial digital syndrome	2	1.70%	-
Ellis van crevland syndrome	1	0.85%	-
Golden harr syndrome.	3	2.60%	-
Multiple congenital anomalies	21	18%	7

Intellectual Disability: Of 82 cases (14.9%) referred for intellectual disability, 2 cases (2.4%) had chromosomal abnormality, with ring chromosome and translocation. Consanguinity is associated with 12 cases (17%).

Infertility: Of 38 cases (6.9%) referred for infertility, 12 cases were male infertility (2.2%) and 26 cases were female infertility (4.7%). Consanguinity was associated with 9 cases (34.6%) of female infertility and 7 cases of male infertility. Among the female infertility, one case showed 9qh+ and another with 46, XY female. Among the male infertility, one case had chromosomal breakage and 4 were azoospermia.

DISCUSSION

Consanguineous marriages have been practiced

since the early existence of modern humans and in several global communities with variable rates depending on religion, culture, and geography [1]. The strengthening of the family relationships is of primary importance in the preference for close kin unions, with economic benefits an additional consideration. [2]. The main impact of consanguinity, however, is an increase in the rate of homozygotes for autosomal recessive genetic disorders. At present, about 20% of world populations live in communities with a preference for consanguineous marriage and 8.5% of all children have CON parents. [5].

The incidence of CON in the literature varies from 2-6% and in India 5-60 % [1]. In the developed countries CON marriage is accounted for 26-34% of perinatal mortality [3,6]. In our study CON marriages was reported in 198 cases (36%). Our finding was similar to Roya M et al who also reported in 37% of cases. [7]. But Rabah et al had reported the highest prevalence of consanguineous marriage (54.4%) [5].

Most frequent type of marriage is the first cousin with the incidence in Islamic countries varies from 10-45% and estimated consanguineous ratio is 30-85%. In the present study, the most common degree of CON marriages was first cousins 51.7% and uncle niece 48.2%. Similar finding was reported by Jain et al who had recorded first cousin marriage 50.6% and uncle niece marriages 42.4% [8]. Another study conducted by Roya et al reported the prevalence of CON marriage among the first cousin was 14.5% and uncle niece was 22.5% which is significantly lower than our finding [7].

Marriage between cousin accounts for over 10% of marriages worldwide [9]. Among CON marriage families, the blood relationship of uncle niece is 22.5% seems to have the highest risk of affecting offspring [3]. CON parents supporting one genetically abnormal child are almost 13 times more likely to give birth to another affected child as compared to non-CON parents [1,5]. Of 550 cases evaluated, chromosomal abnormalities both numerical and structural abnormalities were detected in 54 cases (9.81%). Numerical abnormalities were seen in 38 cases (70.3%) and structural abnormality was seen in 16 cases (29.6%). Consanguinity with chromo-

somal abnormality was seen in 19 cases (3.4%), numerical in 13 cases (68.4%) and structural anomaly seen in 6 cases (31.5%). The study by Hammamy et al stated the incidence of chromosomal abnormality is 0.5-1%, numerical is 50% and structural is 40%. The incidence of a genetic disorder in India is found to be 2.3 %.(1). Another study by Rabah et al showed that the CON marriage is detected in 28.8% of patients with chromosomal disorders including Down's syndrome [5]. Razieh et al and Dubey et al reported the chromosomal abnormality in 5.11% and 12.4% of cases respectively [10,18]. Amudha et al and Muller et al had outlined the effect of CON on chromosomal abnormalities [5,12] Amudha et al added that chromosomal abnormalities occur at postzygotic mitosis or transmitted because of errors at meiosis in parental gametogenesis [12].

In our study 132 couples were referred for Bad obstetric history, CON marriages are present in 54 couples account for 41% of cases referred. First cousin marriage seen in 54.16% and uncle niece marriage accounted for 37.5% of cases, far relative in 8.33% of cases. A Study conducted by Amuda et al reported 42.38% of couples with BOH have consanguineous marriage [12]. Studies by Razieh et al [10], Isa et al [13], and Aruna et al [14] also concluded that the couples with BOH have consanguineous marriage in 46%, 23% and 28.7% of cases respectively.

The mean maternal age of the first conception was 22.2 years, ranging from 15-36 years. Pregnancy loss was reported in 58.9% of cases in the first trimester, 16.6 % in the second trimester and 14.2% in the third trimester. Razieh et al reported mean maternal age is 27.4yrs [10], Warburton reported 26.8yrs as mean maternal age and documented that chromosomal abnormality accounts for 50% of all cases of spontaneous abortion and 60% of first trimester losses [11].

In our study, among the 132 couples of BOH, chromosomal abnormalities was seen in 8 cases (3.03%) of which 3 were males and 5 were females. Out of 8 chromosomal abnormalities, 3 cases (37.5%) were an inversion, 2 cases (25%) were Translocation, and 3 cases (37.5%) were heterochromatic variation. Our results were comparable with that of Usha datta et al who

reported an incidence of chromosomal abnormalities in 3.35% of cases. Various other authors reported an incidence ranging from 1.5% - 12.5% [4,10,16-18].

In our study Down syndrome was seen in 41 cases (7.4%). The chromosomal variations observed were trisomy in 33 cases (80.4%), translocation in 5 cases (12.1%) and normal variants in 3 cases (7%). In literature, the most common chromosomal variation observed in Down syndrome is trisomy 21 [1], with frequency ranging from 87% - 93%. Other common variations are translocation and mosaicism ranging from 2.5% - 8% [19-22]. Anupam k et al stated that the origin of extra chromosome 21 is due to meiotic non disjunction which was found to be 79.2% maternal, 20.6% paternal origin [19].

In our study 82 cases (14.9 %) were referred for intellectual disability (ID). Out of this 12 cases (14.6%) was associated with consanguinity and 2 cases (2.4%) with Chromosomal abnormalities including ring chromosome 22 and inversion in 1 case each. Moghe et al and Velogaleti et al reported an incidence of chromosomal abnormalities in 18.9% and 11.1% of cases ID respectively (25, 26). Globally the reported incidence of CON with intellectual disability in the literature varies from 2 to 60% and in India between 5 to 60% [22]. The most common causes of ID are idiopathic (50%), Genetic (35%), and environmental (15%). Genetic factors associated with ID are chromosomal anomalies, single gene disorder, and syndromes [24]. Consanguineous marriages are associated with higher prevalence of mental retardation [32].

In the present study, Congenital anomalies were seen in 118 cases (21.4%) and CON were noted in 34 cases (28.8%). In India analysis of almost all published cases on Multiple Congenital Anomalies (MCA) showed the occurrence to be 1.94-2.03 % [12]. Congenital anomalies began to emerge as one of the major childhoods health problems and most common etiology are Genetic (30-40%) and Environmental (5-10%) of cases. Genetic factors associated with congenital anomalies are chromosomal anomalies (6%), in which single gene disorder constitutes 25%, multifactorial (20-30%) [27]. Congenital disorders have been reported to be more common among consanguineous progeny with presence

of congenital anomaly in 3.67% of cases and excess level of congenital defects seen in 1st cousin offspring's ranging from 0.7 to 7.5% [9,28,29]. Table 4 shows the prevalence of congenital anomalies in relation to consanguinity. Most common congenital anomalies associated with consanguinity are eye anomalies (13.5%), renal anomalies (6.8%), and Nervous system anomalies (5.9%). Many different types of genetic disorders have been reported to be more common among consanguineous progeny, for example, congenital disorders, including neural tube defects and congenital heart defects [28]. Autosomal recessive hearing loss disorders and visual defects such as early-onset retinal dystrophies, primary congenital glaucoma and anophthalmos also are present at increased prevalence [9,28].

Among the major congenital anomalies CNS and CVS anomalies have been reported frequently and more common in CON progeny and analysis in India showed the occurrence to be 1.94-2.03% [30]. In our study CNS malformations were seen in 7 cases (5.9%), more prevalence in male with 2 cases associated with CON. Kulkarni reported the incidence of neural tube defects seen in 1.1%, higher incidence in CON families, 1.42% when they are 1st cousins, 0.86% when they are far related, 0.57% in non-CON, 2.06% in uncle niece [31]. Consanguinity rates were noted to be higher among parents of newborns with congenital hydrocephalus and neural tube defects than in the general population [33,34].

Cardiac anomalies are seen in 4 cases (3.4%) in our study. A different picture emerges from the large literature on congenital heart defects, which are conservatively estimated to have an incidence of 50/1,000 live births (5%). Although a consistent positive association between consanguinity and disorders such as ventricular septal defect and atrial septal defect has been demonstrated, indicating the involvement of common variants [35]. Bittles reported the incidence of neural tube defects and congenital heart disease are more common in consanguineous progeny [36].

In our study eye anomalies were reported in 16 cases (13.5%) including Retinitis pigmentosa, Retinal dystrophy, congenital lacrimal sinus, congenital ptosis and macular dystrophy and 7

cases (43.8 %) were associated with consanguinity. One case had translocation of chromosome 6 and 12. Bittles reported eye anomalies like early onset retinal dystrophy, anophthalmos, microphthalmos are present at increased prevalence in consanguineous progeny [36].

CON causes congenital defects, single gene disorder including sensori-motor defects, psychological disorders and adult onset diseases and in relation to MCA seen in 3.67% as compared to 1.15% non CON [36]. Hoodfal et al reported that CON marriages have an effect on the rates of reproductive loss, congenital malformation and genetic disorders. Rabia et al studied CON marriages was detected in 80% of Ambiguous genitalia, 72.25 % in Multiple congenital anomalies, and 62.2% of patients with blood disorders [2]. In our study Ambiguous genitalia was reported in 7 cases (5.9%), with CON in 2 cases.

Anupam K et al analysed and reported the incidence of male infertility associated with CON in 7 cases (0.3%) [19]. In our study 38 cases (12 males and 26 females) of infertility were referred. Consanguinity was associated with 9 cases (34.6%) of female infertility and 7 cases of male infertility. Among the female infertility, one case showed 9qh+ and another with 46, XY female. Among the male infertility, one case had chromosomal breakage and 4 were azoospermia.

CONCLUSION

Improving socioeconomic conditions and better access to health care will impact the effects of consanguinity, with a shift from infant and childhood mortality to extended morbidity. At the same time, a range of primarily social factors, including urbanization, improved female education, and smaller family sizes indicate that the global prevalence of consanguineous unions will decline. This shift in marriage patterns will initially result in decreased homozygosity, accompanied by a reduction in the expression of recessive single-gene disorders. (bittlescon human evo)

Conflicts of Interests: None

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