PROJECT REPORT

Estimation of Parental Consanguinity in Children with Communication Disorders

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Abstract

Background: Consanguineous marriage is commonly practiced in the region of North Karnataka (Bittles, 2002). The objectives of this study were to estimate the prevalence of parental consanguinity and establish a pedigree analysis in those with communication disorders and history of parental consanguinity in North West region of Karnataka.

Data Collection: Data were collected by reviewing the clinical records of clients who visited JSS Institute of Speech and Hearing, Dharwad between January 2015 to October 2017. Further, prospective data were collected from clients who visited the institute between November 2017 to October 2018. Additionally, data obtained from free speech and hearing camp organized in Dharwad were included. A total of 2000 individuals below 18 years of age who were diagnosed as having communication disorders were included for the study. Information on parental consanguinity was obtained using a questionnaire.

Results: It was found that among the 2000 individuals with communication disorders, 1257 were males and 743 were females. Further, there were 676 children with communication disorders found to have history of parental consanguinity (33.8%). Among them, 612 of the children had second degree parental consanguinity and 64 of the children had third degree consanguinity. It is also observed that prevalence of parental consanguinity was higher in children with hearing loss compared to other communication disorders.

Conclusions: Prevalence of parental consanguinity in children with communication disorder is high in northern part of Karnataka. The rate of prevalence in the current study is still similar to studies that were conducted 10 years back. This indicates the need for widespread awareness and public education regarding the adverse effect of consanguineous marriages.

CHAPTER -1 INTRODUCTION

Consanguineous marriage is known as a marriage between two people who are closely related. Consanguineous marriages are common in Asian, Middle East and African population. It is reported that approximately one billion of the global population live in communities with a preference for consanguineous marriage (Bittles & Black, 2010; Modell & Darr, 2002). Population which comes under similar beliefs, traditional customs and rituals would prefer consanguineous marriage in terms of keeping property within their family (Rao et al., 2009). High prevalence of consanguineous marriage has been reported in southern part of India especially in the northern districts of Karnataka and Tamilnadu (Bittles, 2002). It is most commonly observed in low socio economic population (Rao et al., 2009).

Consanguineous marriages can cause genetic defects to run in families (Bittles et al., 1991; Hamamy, 2012; Naibkhil & Chitkara, 2016). It is reported that 50 per cent of the genetic make-up is shared between parents and children, brothers and sisters. Similarly, uncle and niece share 25 per cent and first cousins share 12.5 per cent of their inherited genetic material as they originate from a common ancestor (Asendorpf, 2001). In that case if there are any `silent' genetic defects, then such errors manifesting as a disease in the child of a consanguineous parents is high. In contrast, possibility for sharing a defective gene in non-consanguineous partners is extremely rare (Verma, 2000).

Studies have assessed the consequence of consanguineous against non-consanguineous marriage have shown that consanguinity leads to infant mortality before, during or immediately after birth, increased incidence of birth defects, genetic diseases including blinding disorders, blood cancer (acute lymphocytic leukemia), breathing problems for children at birth (apnea), increased susceptibility to disease etc. It is reported that there is high prevalence of communication disorders in parental consanguinity (Bener, Hussain & Teebi, 2007).

National status:

High prevalence of consanguineous marriage has been reported in southern part of India especially in the northern districts of Karnataka and Tamilnadu (Bittles, 2002). A door to door survey was conducted in Shindoli village of District Belgaum of Karnataka to study the prevalence of consanguinity, its effect on fetal loss, obstetric complications, neonatal mortality and congenital anomalies. Results showed 34% prevalence of consanguineous marriage in the targeted population wherein 55% of women belonged to the age group of 15 to 30 years where the majority was illiterates and 74.4% of them were housewives. There were no significant difference between consanguineous and non-consanguineous groups in number of neonatal births and congenital abnormalities (Nath, Patil & Naik, 2004).

Strength of association of family history and consanguinity with permanent hearing impairment in infants was studied by Selvarajan et al (2013). The study was carried out in Chennai, Tamilnadu. In this case-control study there were 420 infants with permanent hearing impairment (control group) and normal hearing sensitivity (experiment group). Parental interview was carried out for both the group to collect the information on family history of hearing impairment and consanguineous marriage. It was found that 18.6% showed family history of hearing loss and consanguinity was seen in (39.5%) of the hearing-impaired group.

Epidemiological study was done on 1076 cases in the age group of 0-14 years attended deaf school for children in and around Hyderabad. For the identified cases with hearing impairment, detailed family history and consanguinity information was obtained. Among 1076 cases with congenital hearing impairment, 41.73% (449) of the cases had a history of parental consanguinity. Further results revealed high rate of consanguinity (44.53%) in cases with non –syndromic deafness (Reddy et al., 2006).

Effect of parental consanguinity and chromosomal abnormality on mental retardation with or without multiple congenital abnormalities has been investigated using retrospective data of clients. Among the 1376 clients information on family history including pedigree over three generation as well as chromosomal analysis were carried out to know significance of consanguinity. Consanguinity was observed in 412 participants (29.94%) and chromosomal abnormality was seen in 626 (45.49%). Consanguinity was significantly associated with mental retardation and or multiple congenital abnormalities with chromosomal abnormality (Rajangam & Devi, 2007).

Retrospective analysis was carried on clients with cleft lip and palate treated in the craniofacial hospital at SDM College of Dental Sciences and Hospital, Dharwad, India. Investigation was carried out to know the association between cleft lip (CL) and palate (CP) and consanguinity. A total of 1247 clients were studied and 47.2% patients' parents had consanguineous marriage. Consanguinity was seen prominently in males (60.2%) and was comparatively lower in female patients (39.7%). Among the 40.9% patients suffered from CL/P, 36.7% and 22.2 % of the individuals were found to have CL and CP separately. Comparatively males had higher incidence of clefts when compared to females. There was a statistically, significant association ($p = 0.04$) between consanguinity and cleft palate (Rajeeve et al., 2017).

A community based Cross sectional study was carried out to find the association between consanguinity and congenital anomalies, autosomal recessive disorders, perinatal and antenatal morbidities. A total of 130 married couples from Kalaburgi, Karnataka, India was taken for the study. Among 130 married couples, 58 couples had consanguineous marriage (44.6%). Among these families, consanguineous marriages were more in Muslim families (56.05%) when compared to Hindu families (25%). Prevalence of abortions and preterm deliveries was noted to be 60% and 64.28% respectively in consanguineous

marriages. It was found that consanguineous marriage was one of the causes resulting in hearing defects (66.6%) in the community.

In cases of individuals with vision deficits, almost all affected individuals belong to consanguineous families and a 100% of all vision defects were seen in consanguineous conceptions. Sakre et al., et al (2017) investigated the association of consanguinity in pediatric neurological disorders. 152 children were recruited for a study from various units of pediatric ward in Velammal medical college hospital, Madurai, Tamilnadu, India. Results showed out of 152 cases 69 (45.3%) of them found to have parental consanguinity. Among cases with parental consanguinity prevalence of hearing impairment was 5 (3.2%), speech delay 2 (1.3%), mental retardation 11 (7.2%), seizure disorder 27 (17.7%), visual impairment 2 (1.3%) and ataxia telangiectasia 2 (1.3%). It was also observed that prevalence of neurological disorders were slightly higher in cases with parental consanguinity (Maheswari &Wadhwa , 2016).

International status:

It has been reported that risk for congenital/genetic disorders is most noticeable for autosomal recessive disorders and depends on the degree of relatedness of the parents (Teeuv et al., 2010). Based on a retrospective analysis carried out in Omani children (Khabori & Patton, 2008) it was found that majority (70%) of the deaf children were from parents of consanguineous marriages, whereas only 30% of them were from non-consanguineous unions. In those with consanguineous marriages 70.16% were first cousin marriages, 17.54% were second cousins, and 10.86% were from the same tribe. The proportion of the first cousin marriages was higher than the background rate of first cousin marriages in Oman. In the total cohort, 45% had other family members with hearing loss. The chance for being affected in the consanguineous group was higher than the non-consanguineous group (29.7% versus

15.3%). In most cases the affected relative was a deaf sibling (67.8%). A higher rate of consanguinity was demonstrated amongst parents of deaf children in Oman and is associated with a higher frequency of autosomal recessive deafness..

Study was done to delineate the role of consanguinity on congenital malformations in Khominishahr rural population, Isfahan, Iran. There were 518 malformed population (case group) and 518 normal subjects (control group) who were randomly selected from khominishahr rural population. The results revealed frequency of consanguinity of parent's was 59.7% in clinical group and 31.5% in control group. This difference was statistically significant ($p < 0.001$) as reported by Kushki and Zeyghami (2005).

Study was carried out by El-Din and Hamed (2008) to investigate the prevalence of sensorineural hearing loss in offspring's of consanguineous marriage at Medical Genetics Center, Ain Shams University, Egypt. Study was carried out on 950 children with congenital hearing loss and results showed 71.2% of hearing impaired children had history of parental consanguinity among them 47.3% were first cousin ,36% were second cousin and 16% had remote consanguinity. Further they reported 44.2% of cases had severe degree of sensorineural hearing loss (71-90 dBHL), 24.3% had profound hearing loss (>90 dBHL). Musani et al.(2011) studied the frequency and causes of hearing impairment in patients attending the outpatient department of Civil Hospital Karachi. A total of 600 patients with a complaint of hearing impairment were considered for the study. It was found that the frequency of conductive hearing loss was 50%, sensorineural hearing loss 20% and mixed hearing loss in 30%. CSOM remains the most common cause in this study. In majority of cases of sensorineural hearing loss, prenatal and perinatal factors predominate. Consanguinity was the most frequent factor in those with sensorineural type of hearing loss.

Durkin et al (1998) estimated the prevalence of mental retardation and risk factors associated with the same. A total of 6365 children was screened for disability using questionaries' further for children who failed in questionnaires' detailed medical and psychological evaluation was carried out. Results revealed that 19.0/1000 had serious retardation, 65.31/1000 of children had mild retardation. Lack of maternal education was strongly associated with serious and mild retardation other factors that were independently associated includes history of prenatal difficulty, neonatal infection, post brain infection, traumatic injury and malnourishment. Further research need to be assessing the contribution of consanguineous marriage.

Bangash, Hanafi andIdrees (2014) conducted a pilot cross-sectional study to determine the risk factors associated with cerebral palsy (CP). Data were collected through an interview questionnaires' from the mother of CP children from a population of 600. Results revealed that major risk factors identified were home and assisted delivery 5 (75%), consanguinity 10 (50%), infection 8 (40%) and lack of antenatal care 6 (30%). Lotfi (2004) studied 1352 infants and preschool aged children to establish prevalence of SNHL due to consanguineous marriage in first cousin and second cousins. Consanguinity was found among 45.7 percent in first cousin and 17.2 percent in second cousin. Hereditary Factors were thought to be the cause of 863 (62.9 percent) of bilateral SNHL children in this research.

Zakzouk, Sayad and Bafaqeesh (1993) conducted a random sample survey in Saudi infants and children to understand the prevalence of consanguineous marriage and its effect on the prevalence of hereditary sensorineural hearing loss. Parents of 21.1% of the children studied had first cousin consanguineous marriage and second cousin consanguinity was present in 23%. The overall prevalence of hereditary sensorineural hearing loss was 1.7%. A higher prevalence of 2.8% of this type of deafness of more distant consanguinity and 1.4% among non-consanguineous family's children. Bener et al., 2005 studied frequency of hearing

loss and its association with consanguinity among Qatari population. Correlation between hearing loss and Rhesus (Rh) blood groups were also investigated. Results revealed more common parental consanguinity among individuals with hearing loss when compared with individuals with normal hearing. There was a strong correlation between hearing loss and consanguinity which were influenced by parental literacy level and blood group of parents.

Recent researches in Qatar confirmed a high homogeneity and level of inbreeding in Qatari hereditary hearing loss HHL patients. (Giratto et al.,2014) Among all HHL causing genes, GJB2, the major player worldwide, accounts for a minor proportion of cases and at last 3(LOXHD1, *MYO15A* &*BDP1)* additional genes have been found to be mutated in Qatari patients. Interestingly, one gene, BDP1, has been described to cause HHL only in this country. These results point towards an unexpected level of genetic heterogeneity despite the high level of inbreeding. Rabia and Maroon (2005 studied the effect of consanguineous marriage on reading disability investigation was carried out between reading disability children and normal reading young children. Results indicated the rate of reading disability among children of first cousin was high compare to second cousin ,distant related parents or unrelated parents .

From the above findings it can be illustrate that, high prevalence of communication disorders observed in offspring's of consanguineous couples compare to non-consanguineous couples.

Need for the study

Studies have proven the association between consanguinity and birth defects. It is well studied that prevalence of hearing impairment is high in children with a history of

parental consanguinity (Zakzouk, 2002; Zakzouk, Sayad & Bafaqeesh 1993; Bener et al. 2005). Further, consanguinity is a renowned risk factor for genetic disorders including elevated levels of global developmental delay and mild and severe intellectual and developmental disabilities (Saad et al. 2014). High prevalence of speech, language and hearing problems in children with history of parental consanguinity urges the need for estimating its prevalence in different communities. High prevalence of consanguineous marriage has been reported in southern part of India especially in the northern districts of Karnataka (Bittles, 2002). Though various studies have estimated the prevalence of consanguinity in northern Karnataka (Nath, Patil & Naik, 2004; Bittles, 2002) its frequency of occurrence is not estimated in Dharwad.

Henceforth, it is important to establish prevalence rate of parental consanguinity in children with communication disorders for a specific region. These findings will be essential in creating awareness on prevention, early identification and consequences of consaguinous marriage. It is observed that there are no recent studies on consanguinity and communication disorders in this geographical area. Previous studies were conducted almost 2 decades back. Hence, there is a need to investigate the prevalence of consanguinity among those with communication disorders. Such an investigation may give an estimate of change in prevalence of consanguinity and communication disorders over a period.

Aim of the study

The aim of the study was to estimate the prevalence of parental consanguinity in the region of North western Karnataka, Dharwad.

Objectives of the study

1. To identify children with communication disorders and history of parental consanguinity using a questionnaire (Basavaraj, Savithri, Manjula & Sudharshan, 2017). (Communication disorders included are mentioned in Appendix D)

2. To estimate parental consanguinity in children diagnosed with communication disorders

3. To establish a pedigree analysis in those with communication disorders and history of parental consanguinity.

CHAPTER -2 METHODS

The study aimed to estimate the prevalence of parental consanguinity in children with communication disorders in North western district of Karnataka, Dharwad. Children with communication disorders and history of parental consanguinity were identified using a questionnaire (Basavaraj, Savithri, Manjual & Sugharshan, 2017). The study also established a pedigree analysis in those with communication disorders and history of parental consanguinity.

Participants

There were 2000 clients below the age of 18 years diagnosed with any communication disorder were involved in the study. Among them 1257 were males and 743 were females. Gender wise distribution of participants can be seen in Table 2.1. All the participants were native speakers of Kannada and were residents of Dharwad district. The clients chosen for the study include those identified from a retrospective analysis of patients visited JSS Institute of speech and Hearing, Dharwad over a period of 3 years (January 2015 - October 2017). Additionally, clients who visited the institute between November 2017 to October 2018 were considered. Further, clients who attended free speech and hearing camps organized at Dharwad were also included. The entire study was executed adhering to the 'Ethical guidelines for bio-behavioural research involving human subjects' of the All India Institute of Speech and Hearing (2009).

Table 2.1: Distribution of children with communication disorder with respect to gender.

	Total Clients with communication disorder	Male Clients with communication disorder	Female Clients with communication disorder
Retrospective data	958	619	339
Prospective data	816	524	292
Camp data	226	114	112

Procedure:

The study was carried out in 3 phases. In the phase 1 screening questionnaires on communication disorders and parental consanguinity was administered. In the Phase 2, diagnosis of communication disorders was carried out. Pedigree analysis was carried out in the phase 3. However, information on pedigree was not obtained in the retrospective data.

Phase 1: Administering questionnaire

In this phase a screening questionnaires on communication disorder and parental consanguinity was administered (Basavaraj ,Savithri, Manjula and Sudharshan, 2017). The checklist has two sections. First section consists of 16 questions to identify speech, language and auditory disorders in children. The second section consist 37 questions to identify the high risk factors based on birth history and family history and parental consanguinity in children with communication disorders.

The questionnaire was administered on clients visited the OPD of the institute as well as those who attended the camp. The questionnaire was administered to parents or caretakers

of children with a complaint of speech, language and hearing problems. Patients who were identified having a communication disorder in the questionnaire were subjected to detailed speech, language and audiological evaluations and were diagnosed according to DSM-5 and ICD 10 criteria by a qualified Speech Language Pathologist and Audiologist. Prior administering the questionnaire each parent was explained about the study and a written consent was taken.

Phase 2: Diagnostic evaluation:

The clients identified in phase 1 were subjected to detailed speech, language and hearing evaluation. In order to diagnose speech and language disorders, standard test materials such as Receptive Emergent Expressive Language Scale (REELS), Receptive Expressive Language Test (RELT), and 3 Dimension Language Acquisition Test (3D LAT), Stuttering Severity Instrument (SSI), Early Reading Skills (ERS) were used .

Routine audiological evaluation was carried out on all the patients with complaint of hearing problems. Pure-tone air conduction and bone conduction thresholds were obtained at octave frequency between 250 Hz to 8000 Hz and between 250 Hz to 4000 Hz respectively using modified version of Hughson and Westlake procedure (Carhart & Jerger, 1959). Further, speech recognition threshold and speech identification score were obtained using appropriate test materials. Middle ear status was evaluated by varying the pressure from +200 dapa to -400 dapa for probe tone frequency of 226 Hz. Further, Ipsilateral and contralateral acoustic reflex thresholds were measured for 500 Hz, 1000 Hz, 2000 Hz and 4000 Hz pure tones. Hearing thresholds in pediatric population was confirmed using Auditory Brainstem Response (ABR) recorded for click /tone bust stimuli. Further, Transient evoked Oto acoustic emissions/Distortion product Oto acoustic emissions were recorded using Otodynamics ILO 292. Presence of OAEs were determined based on Signal to Noise Ratio (SNR) of + 6 dB in

three consecutive frequencies with reproducibility greater than 75%. Patients having average audiometric threshold at 500, 1000, 2000 & 4000 Hz greater than 26 dB HL were considered as hearing impaired (WHO, 2008) and were included in the study.

Instrumentation

Calibrated diagnostic audiometers (Interacoustics AD-629 or ALPS AD 2000) were used to perform air conduction and bone conduction thresholds and speech audiometry. For Speech audiometry, Phonetically balanced (PB) word list in Kannada and Hindi were used which were developed by Yatiraj and Vijayalakshmi (2005) and Abrol et al (1972) respectively. Middle ear status was evaluated using a calibrated immittance meter (Interacoustic AT-235). Further, ABR and OAE were performed using Interacoustics Eclipse-15 and ILO DP Echoport systems respectively.

Phase 3: Pedigree analysis:

Individuals with consanguinity and communication disorders were followed up to perform a pedigree analysis. The members of the family who are affected by a genetic trait were noted in the pedigree display . Pedigree analysis was implemented by using the MENDEL packagae (Lange et al. 1988). A minimum of four generations were considered for the pedigree analysis. This was to identify the presence of any autosomal recessive transmission in their families. Pedigree chart and respective details were further analyzed by a Geneticist.

Analysis

Data obtained in the retrospective and prospective study were analyzed separately. Data obtained from the clients were tabulated and analyzed using SPSS software. Descriptive analysis was carried out to establish the prevalence of consanguinity in children with communication disorder.

CHAPTER -3 RESULTS

The aim of the research project was to estimate the prevalence of parental consanguinity in children with communication disorders in the north western district of Karnataka, Dharwad. Retrospective and prospective data collected from the patients visited JSS Institute of Speech and Hearing, Dharwad as well as clients who attended speech and hearing camp at Dharwad were analyzed. The results obtained are described under three divisions with reference to the objectives of the study.

1. Identification of children with communication disorder and history of parental consanguinity

A total of 816 children who visited JSS Institute Speech and Hearing, Dharwad between November 2017 and October 2018 were screened to identify any communication disorder as well as parental consanguinity. Further, 226 clients who attended free speech and hearing camps organized at Dharwad were also subjected to the screening questionnaire. There were 1042 children who failed in the questionnaire and were suspected to have communication disorders. Among them, 638 were males and 404 were females. Additionally there were 958 patients diagnosed with a communication disorder were identified from the retrospective analysis of data of clients who visited JSS Institute of speech and Hearing, Dharwad over a period of 3 years (January 2015 - October 2017). A screening questionnaire developed by Basavaraj, Savithri, Manjula and Sudharshan, (2017) was used. Distribution of data obtained from the retrospective, prospective analysis and camp are seen in Figure 3.1.

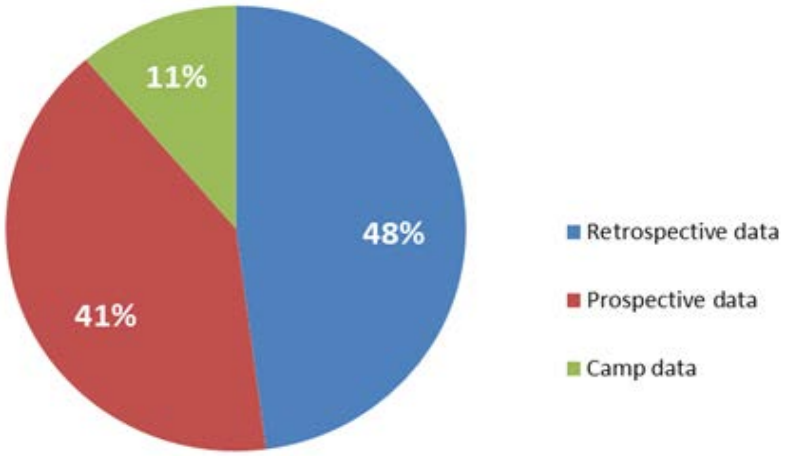

*Figure 3.1:*Distrubution of data of children with communication disorders.

2. Estimation of parental consanguinity in children with communication disorders.

Analyzing the retrospective, prospective and camp data there were a total of 2000 children diagnosed to have communication disorders. It can be seen in figure 3.2 that among the diagnosed clients, 1257 (63.75%)were males 743 (37.15%) were females.

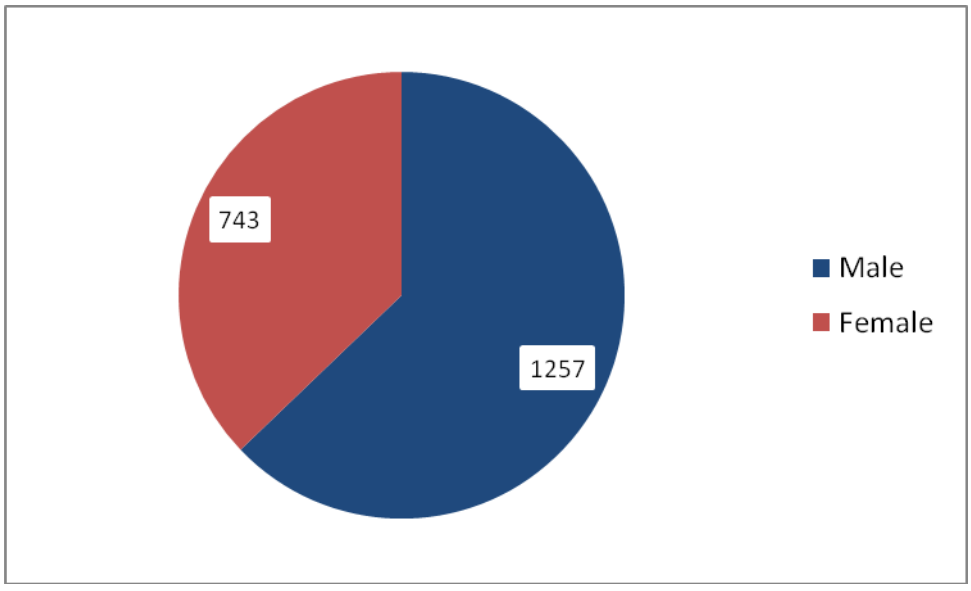

Figure 3.2:Number of clients having communication disorders with respect to gender

Number of clients with and without history of parental consanguinity are given in Figure 3.3.

There were 676 (33.8%) of children with communication disorders had a history of parental consanguinity. However, 1324 (66.2%) of children with communication disorders did not have history of parental consanguinity

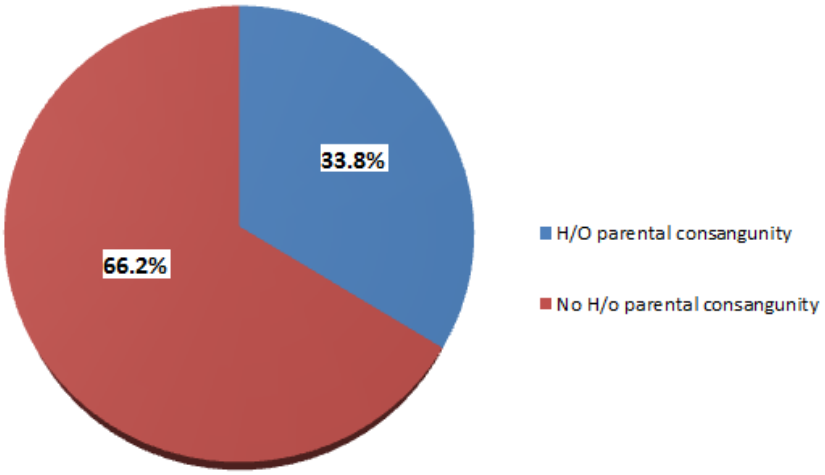

Figure 3.3 : Number of children with communication disorders with and without history of parental consanguinity.

Prevalence of parental consanguinity was calculated using the below mentioned formula

$$\text{Prevalence} = \frac{\text{Number of children with history of parental consanguinity}}{\text{Total number of children with communication disorders}}$$

It was found that prevalence rate of parental consanguinity was 676 (33.8%).

Among those clients, there were 612 (90.53%) clients had second degree parental consanguinity, 64 (9.46%) clients had third degree of parental consanguinity as given in Figure 3.4 higher number of male children had history of parental consanguinity compared to female children (Male: 402 (59.46%), Female :274(40.53%) as given in Figure 3.5.

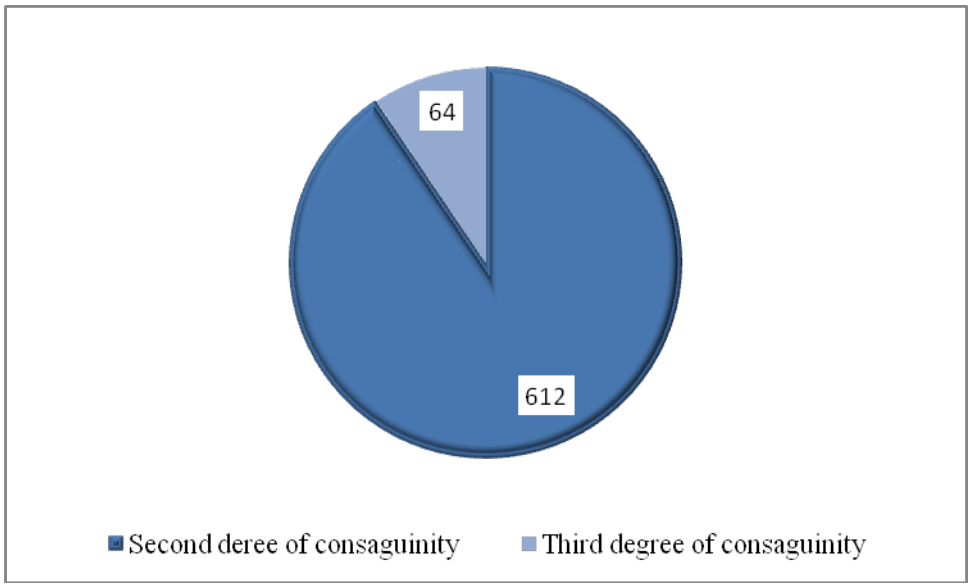

*Figure 3.4:*Number of clients with history of parental consanguinity with respect to degree of consanguinity.

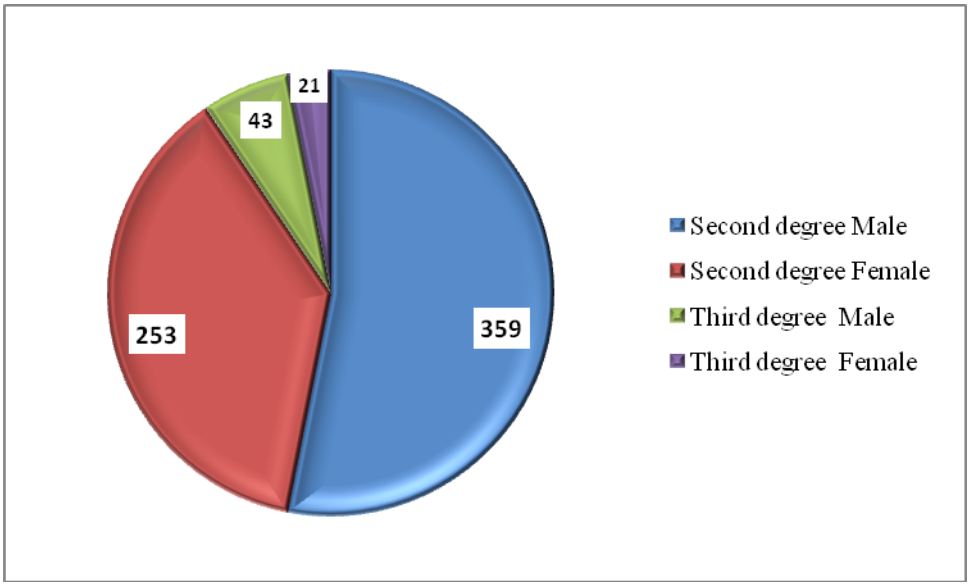

*Figure 3.5:*Number of clients with history of parental consanguinity with respect to gender.

It is observed in Table 3.1 that prevalence of parental consanguinity was higher in clients with hearing loss compared to other communication disorders. There were 465 clients with hearing impairment having either second or third degree of consanguinity. Intellectual disability, cerebral palsy and specific language impairment were also found to be associated with history of parental consanguinity. Followed by other childhood speech and language disorders.

*Table 3.1***:** Distribution of communication disorder in patients with history of parental consanguinity

Communication disorders	**Total**
DSL Secondary to HL	465
DSL Secondary toID	57
DSL Secondary to CP	56
SLI	55
SSD	15
DSL Secondary to ASD	9
DSLSecondary to SD	8
FD	4
DSL Secondary to Synd	4
DSL Secondary to CLP	2
LD	1

Note:

DSL secondary to HL: Delayed speech and language secondary to hearing impairment

DSL secondary toID: Delayed speech and language secondary to intellectual disability

SLI: Specific language impairment

DSLsecondary to CP: Delayed speech and language secondary to cerebral palsy

SSD: Speech sound disorder

DSL secondary toASD: Delayed speech and language secondary to Autism spectrum disorder

DSLsecondary toSD: Delayed speech and language secondary to seizure disorder

FD: Fluency disorder

LD: Learning disability

DSL secondary to CLP: Delayed speech and language secondary to Cleft lip and palate

DSL secondary toSynd: Delayed speech and language secondary to syndrome.

3. Pedigree analysis in those with communication disorders and history of parental consanguinity.

The study examined a total of 209 pedigrees of children with communication disorder with history of parental consanguinity. The analysis showed 183 clients showed history of recessive gene, 10 clients had autosomal dominant inheritance, 8 had cytoplasmic gene and 8 had holandric gene. With reference to result obtained it can be inferred that highest number of clients had recessive inheritance. Examples of different inheritance obtained from the pedigree analysis are given in the appendix A.

CHAPTER - 4 DISCUSSIONS

Main objective of the project was to estimate the prevalence of parental consanguinity in children with communication disorders in the region of North Karnataka, Dharwad district. The results of the study revealed that out of 2000 patients studied 676 (33.8%) were found to have history of parental consanguinity. The findings are in consonance with the studies carried out in north Karnataka region. A study by Nath ,Patil & Naik (2004) in the region Belgaum showed the prevalence rate of parental consanguinity of 36% in hearing impaired population which is similar to the findings of the present study . Similarly, a study by Rajeev et al., (2017) in the region of Dharwad also showed 47.2% of children with cleft lip and palate found to have history of parental consanguinity. Similarly, in region of Kalaburagi, a district of North Karnataka, estimated the prevalence of consanguineous marriage among 130 families was found to be 44.6%. Further, they reported 66.6% of hearing impaired in the community was in consanguineous families. The findings of the study clearly shows that the tradition of consanguineous marriage still persist in the north Karnataka Region. Increased prevalence shows the lack of awareness about risks of having consanguineous marriage in this region of Karnataka.

There is a higher prevalence of consanguineous marriages in Southern region of India compared to its rest. Rao and Narayana (1976) reported 30.3%.of the children had history of parental consanguinity among children with intellectual disability. A study by Reddy et al. (2006) in the region of Hyderabad, Andhra Pradesh identified 1076 children having risk factors causing hearing impairment. Consanguinity was found as a high risk factor in 41.73% (449) of the children. A study by Saleem ,Shankar & Sabeetha (2016) in the region of Tamilnadu also reported a prevalence of consanguineous marriage in 39.2% of children and found positive association between the consanguinity and congenital abnormality. Case control study by Selvarajan et al. (2013) investigated the association between consanguinity,

family history and permanent hearing impairment in infants. Results showed 39.5% of infants had history of consanguinity and 18.9% of infants with family history of hearing loss.

The findings of the study revealed that there is an increased prevalence of consanguineous marriage in northern Karnataka compared to its south. This could be because the tradition of consanguineous marriage that is common in northern part of Karnataka compared to its south. The results highlight the need for increasing awareness in this region using through public education activities.

Prevalence of communication disorders was found more in males compared to females. Similar findings were obtained by studies conducted by Devadiga et al .,2014 ; Shanbal et al.,2015., Konadath et al.,2013; Sinha et al.,2017; Lin et al.,2011; Beria et al.,2007; Kinnon, Leod & Reilly (2007). This could be because parental consanguinity, a recessive type of inheritance observed more in males compared to females. It could also be that the males in India receive more attention and so are better attended for deviation in health status compared to females (Bittles, 2001).

Among communication disorders prevalence of hearing impairment was high in children with parental consanguinity. This shows that hearing impairment was the communication disorder that was reported maximally in this region. This could be due to the symptoms of hearing impairment that are easily identifiable. Moreover, widespread awareness on facilities available for individuals with hearing impairment would have also lead to this. Also, auditory disorders are usually associated with evident signs and symptoms for which medical help is sought and hence patients tend to report to an ENT specialist or general physiscian. In contrast, speech and language disorders are less reported due to the lack awareness on professional services available (Devadiga et al .,2014). The present study findings are in consonance with the study carried out in north Karnataka region (Belgaum) (Nath ,Patil & Naik ,2004) which showed hearing impairment was third highest disability

(7.24%) seen followed by mental disability (5.92%) and speech disability (95.26%). In the region of Kaluburgi (2011) the disability percentage of hearing is about (24.07%), speech (5.5%) and intellectual disability was (6.12%). Similar findings are also observed in south Indian region studies Maheshwari & Wadhwa (2016) reported in the region of Tamilnadu the prevalence of hearing impairment 5 (3.2%) speech delay 2 (1.3%) and intellectual disability 11 (7.2%) among the children with history of parental consanguinity. Reddy et al(2006) in the region of Hyderabad reported among 1076 children 449% of the children were identified to have history of parental consanguinity and (44.53%) were hearing impaired .

Highest number of recessive type of inheritance observed among children with communication disorders with history of parental consanguinity. It could be because in children with parental consanguinity recessive type of inheritance is observed more common and is more prevalent in males compared to females (Bittles et al.,1991; Bittles & Black ,2010; Hamamy et al. 2012; Tadmouri et al. 2009).

CHAPTER 5 - SUMMARY AND CONCLUSION

Consanguineous marriages are common in Asian, Middle East and African population. It is reported that one billion of the world population live in communities with a preference for consanguineous marriage (Bittles & Black, 2010; Modell & Darr, 2002). High prevalence of consanguineous marriage has been reported in southern part of India especially in the northern districts of Karnataka and Tamilnadu (Bittles, 2002). Consanguineous marriages can cause genetic defects to run in families. Studies have assessed the consequence of consanguineous against non-consanguineous marriage have shown that consanguinity leads to death of infants before, during or immediately after birth, increased incidence of birth defects, genetic diseases including blinding disorders, blood cancer, breathing problems for children at birth, increased susceptibility to disease etc. It is reported that there is high prevalence of communication disorders in children with history of parental consanguinity (Abdulbari, Rafat & Ahmad, 2007). Thus the current study was taken up with the purpose to

To identify children with communication disorders and history of parental consanguinity using questionnaires (Basavaraj, Savithri, Manjula & Sudharshan, 2017).

To estimate parental consanguinity in children diagnosed with communication disorders

To establish a pedigree analysis in those with communication disorders and history of parental consanguinity.

To study the above mentioned objectives, 2000 clients below the age of 18 years diagnosed with any communication disorder were involved in the study. The clients chosen for the study include those identified from a retrospective analysis of patients visited JSS Institute of speech and Hearing, Dharwad over a period of 3 years (January 2015 - October 2017). Further, clients who visited the institute between November 2017 to October 2018 were considered. Additionally, the clients attended free speech and hearing camps organized at Dharwad were included. Screening questionnaires on communication disorders and

parental consanguinity were administered (Basavaraj, Savithri, Manjula & Sudharshan, 2017). Those clients identified using the questionnaires' were further evaluated for speech, language and hearing. In order to diagnose speech and language disorders, standard test materials were used and for clients with complaint of hearing problems routine audiological evaluation was carried out. Patients having average audiometric threshold at 500, 1000, 2000 & 4000 Hz greater than 26 dB HL were considered as hearing impaired (WHO, 2008). Further for Individuals with consanguinity and communication disorder, pedigree analysis was obtained. This was to identify the presence of any autosomal recessive transmission in their families. Data obtained in the retrospective and prospective study were analyzed separately. Descriptive analysis was carried out using SPSS software to establish the prevalence of consanguinity in children with communication disorder.

Among the 2000 children diagnosed to have communication disorders,1257(63.75%) were males 743(37.15%) were females. It was found that prevalence rate of parental consanguinity was 676 (33.8%). Among these clients, there were 612 (90.53%) clients had second degree parental consanguinity, 64 (9.46%) clients had third degree of parental consanguinity. Higher number of males 402 (59.46%), children had history of parental consanguinity compared to females 274 (40.53%) children. It is observed that prevalence of parental consanguinity was higher in clients with hearing loss compared (465) to other communication disorders. Intellectual disability, cerebral palsy and specific language impairment were also found to be associated with history of parental consanguinity. Followed by other childhood speech and language disorders.Further, pedigree analysis in 209 clients showed history of recessive gene in 193 clients, 8 had cytoplasmic gene and 8 had holandric gene. It was noticed that recessive type of inheritance was highly common in children with communication disorders with history of parental consanguinity. The study shows increased prevalence of parental consanguinity in children with communication

disorders in northern Karnataka. Hence, the study highlights the importance of public education regarding the negative impacts of consanguineous marriage in this region.

Limitations of the study

Unequal distribution of various speech and language disorders might have led to poor understanding of prevalence of such disorders like Cleft lip and palate, Learning disability, autism, fluency disorders etc. Information regarding socio-economic status, literacy, religion of the parents would have given a better piture on prevalence of consanguinity.

Implications

The present study is an attempt to establish region specific prevalence rate of parental consanguinity in children with communication disorders. The results revealed that prevalence rate of parental consanguinity in this region remains the same compared to findings observed in studies conducted 10 years back. These highlight need for public education regarding the consequences of consanguineous marriage. The study also highlights the importance of awareness programs in general public and other professionals about causes, prevention, identification and rehabilitation of communication disorders.

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APPENDIX

Appendix -A

ಅಖಿಲಭಾರತವಾಕ್ಶ್ರವಣಸಂಸ್ಥೆ, ಮೈಸೂರು-570006

ಮನೆಮನೆಯಸಮೀಕ್ಷೆ

ಸಮೀಕ್ಷೆಯನ್ನುಮಾಡುತ್ತಿರುವವ್ಯಕ್ತಿಯಹೆಸರು:----------

ದಿನಾಂಕ:----------

ವಿಳಾಸ:----------

-

ಗ್ರಾಮದಹೆಸರು:----------

ನೋಂದಣಿಸಂಖ್ಯೆ :----------

ಸಮೀಕ್ಷೆಮಾಡುವಾಗಅನುಸರಿಸಬೇಕಾದಸೂಚನೆಗಳು:

ಪ್ರತಿಪ್ರಶ್ನೆಗೂಹೌದುಅಥವಾಇಲ್ಲಎಂದುಮೂಲಕಉತ್ತರಿಸಬೇಕು.

ಪ್ರಶ್ನಾವಳಿ

1.	ಶಸ್ತ್ರ ಚಿಕಿತ್ಸೆ ಮಾಡಿ ಧ್ವನಿ ಪೆಟ್ಟಿಗೆಯನ್ನು ತೆಗೆದಿದೆ.	ಹೌದು/ ಇಲ್ಲ
2.	ಧ್ವನಿಯು ಕರ್ಕಶವಾಗಿದೆ/ಗೊಗ್ಗರಾಗಿದೆ/ಮೂಗಿನಿಂದ ಮಾತನಾಡಿದಂತೆ ಭಾಸವಾಗುತ್ತದೆ	ಹೌದು/ ಇಲ್ಲ
3.	ಆಹಾರವನ್ನುಅಥವಾನೀರನ್ನುನುಂಗುವಾಗಗಂಟಲಿನಲ್ಲಿನೋವು/ಕಿರಿಕಿರಿ ಉಂಟಾಗುವುದು	ಹೌದು/ ಇಲ್ಲ
4.	ಗಂಡಸುಹೆಂಗಸಿನಂತೆಅಥವಹೆಂಗಸುಗಂಡಸಿನಂತೆಮಾತನಾಡಿದಂತೆಭಾಸವಾಗುತ್ತದೆ	ಹೌದು/ ಇಲ್ಲ
5.	ಅತಿ ಜೋರಾಗಿ ಕೂಗುವ/ಮಾತನಾಡುವ ಅಭ್ಯಾಸವಿದೆ	ಹೌದು/ ಇಲ್ಲ
6.	ಮಗುವಿನಲ್ಲಿ ಮಾತು ಹಾಗು ಭಾಷೆಯ ಬೆಳವಣಿಗೆಯು ನಿಧಾನವಾಗಿರುವುದು	ಹೌದು/ ಇಲ್ಲ

7.	ವ್ಯಕ್ತಿಯ ಬುದ್ಧಿಶಕ್ತಿ ಸಮವಯಸ್ಕರಂತೆ ಇಲ್ಲ	ಹೌದು/ ಇಲ್ಲ
8.	ಮಾತುಅಸ್ಪಷ್ಟವಾಗಿಹೊರಡುವುದು/ಅಕ್ಷರಗಳಉಚ್ಚಾರಣೆತಪ್ಪಾಗಿದೆಉದಾ: 'ಸ' ಅಕ್ಷರದ ಬದಲಿಗೆ 'ತ' ಅಕ್ಷರವನ್ನು ಹೇಳುವುದು	ಹೌದು/ ಇಲ್ಲ
9.	ದೇಹದ ಬೆಳವಣಿಗೆಯು ವಯಸ್ಸಿಗೆ ತಕ್ಕಂತೆ ಇಲ್ಲ/ದೇಹದ ಬಲ ಅಥವಾ ಎಡ ಭಾಗವು ದೌರ್ಬಲ್ಯವಾಗಿರುವುದು (ಲಕ್ವ ಹೊಡೆದಿರುವುದು) ಉದಾ: ಕತ್ತು ನಿಲ್ಲುವುದು, ಕುಳಿತುಕೊಳ್ಳುವುದು, ನಡೆಯುವುದು ಇತ್ಯಾದಿ.	ಹೌದು/ ಇಲ್ಲ
10.	ನಾಲಿಗೆ, ತುಟಿ ಹಾಗು ದವಡೆಗಳ ಚಲನೆಗಳು ಸರಿಯಾಗಿಲ್ಲದಿರುವುದು/ ಬಾಯಿ ಹಾಗೂ ಮುಖದ ಸ್ನಾಯುಗಳು ನಿಶಕ್ತವಾಗುವುದು/ಜೊಲ್ಲು ಸುರಿಯುವುದು	ಹೌದು/ ಇಲ್ಲ
11.	ತುಟಿ ಅಥವಾ ಅಂಗಳಿನಲ್ಲಿ ಸೀಳು ಇರುವುದು/ಹಲ್ಲುಗಳು ವಕ್ರವಾಗಿರುವುದು	ಹೌದು/ ಇಲ್ಲ
12.	ಮಗುವುಗೆ ಮೂರು ವರ್ಷದ ವಯಸ್ಸಾದರೂ, ಪೂರ್ಣ ವಾಕ್ಯದಲ್ಲಿ ಮಾತನಾಡುವುದಿಲ್ಲ/ವ್ಯಾಕರಣಪೂರಿತವಾದ ವಾಕ್ಯಗಳನ್ನು ಬಳಸುವಲ್ಲಿ ತೊಂದರೆ ಇರುವುದು/ ಹೇಳಬೇಕಾದ ಪದಕ್ಕೆ ಮತ್ತೊಂದು ಪದವನ್ನು ಉಪಯೋಗಿಸುವುದು	ಹೌದು/ ಇಲ್ಲ
13.	ಒಂದು ಶಬ್ಧವನ್ನು/ ಅಕ್ಷ್ಯವನ್ನು ಪದೇ ಪದೇ ಹೇಳುವುದು/ಹಿಡಿದು ಹಿಡಿದು/ ಎಳೆದು ಎಳೆದು ಮಾತನಾಡುವುದು ಉದಾ: ನಿ...ನಿ...ನಿ..ನಿಮ್ಮ ಹೆ..ಹೆ..ಹೆಸರು ಏನು? ನನನನನಗೆಗೆಗೆ ಊಟ ಬೇಕು. ಮಾತನಾಡಲು ಹಿಂಜರಿಯುವುದು (ಮಾತಿನ ಬದಲು ಸನ್ನೆಯನ್ನು ಜಾಸ್ತಿ ಬಳಸುವುದು)	ಹೌದು/ ಇಲ್ಲ
14.	ಮಗುವು ಇತರರೊಡನೆ ಸರಾಗವಾಗಿ ಬೆರೆಯುವುದಿಲ್ಲ/ಶಬ್ದಗಳಿಗೆ/ ದೃಶ್ಯಗಳಿಗೆ/ಸ್ಪರ್ಶಕ್ಕೆ ಸರಿಯಾಗಿ ಪ್ರತಿಕ್ರಯಿಸುವುದಿಲ್ಲ/ಒಂದೇ ತರಹ ಇರಲು ಬಯಸುವುದು. ಉದಾ: ಇಡಿ ವಾರ ಒಂದೇ ಬಣ್ಣದ ಬಟ್ಟೆಯನ್ನು ತೊಡಲು ಇಚ್ಚಿಸುವುದು	ಹೌದು/ ಇಲ್ಲ

ಅಖಿಲಭಾರತವಾಕ್ಶ್ರವಣ ಸಂಸ್ಥೆ, ಮೈಸೂರು-570006

ಅಪಾಯದ ಅಂಚಿನ ಅಂಶಗಳ ಕೈಪಿಡಿ

Appendix -B

(ಸಾಮಾನ್ಯಜನರುಮಕ್ಕಳಲ್ಲಿನ ಸಂವಹನ ನ್ಯೂನತೆಗಳನ್ನುಗುರುತಿಸುವಸಲುವಾಗಿ)

ಈ ಕೈಪಿಡಿಯನ್ನುಉಪಯೋಗಿಸಲುಕೆಲವುಸೂಚನೆಗಳು:

- ಮುಂದೆ ಮಾತು ಮತ್ತು ಶ್ರವಣ ದೋಷ ಬರಬಹುದಾದಂತಹ ಸೂಚನೆಗಳನ್ನು ತಿಳಿದುಕೊಳ್ಳಲು, ಮಗುವಿನ ತಂದೆ ತಾಯಿ ಅಥವಾ ಪೋಷಕಇಗೆ ಈ ಕೆಳಕಂಡ ಪ್ರಶ್ನೆಗಳನ್ನು ಕೇಲಬೇಕು.
- ವಯಸ್ಸಿಗೆಅನುಗುಣವಾಗಿ, ಈ ಕೈಪಿಡಿಯಲ್ಲಿ ಸೂಕ್ತವಾದ ವಿಭಾಗದಿಂದ ಆಯ್ದ ಪ್ರಶ್ನೆಗಳನ್ನು ಕೇಳಬೇಕು.
- ಯಾವುದಾದರು ಅಪಾಯಕಾರಿ ಅಂಶಗಲು ಇದ್ದಲ್ಲಿ, ಅವುಗಳು ಇದೆಯೇ ಅಥವಾ ಇಲ್ಲವೆ ಎಂದು ವಯಸ್ಸಿಗೆ ತಕ್ಕಂತೆ ನಮೂದಿಸಬೇಕು

ಹುಟ್ಟಿನಿಂದ ಒಂದು ತಿಂಗಳವರೆಗೆ

	ಅಪಾಯಕಾರಕ ಅಂಶಗಳು	
1.	ಮಗುವಿನ ತಂದೆ ತಾಯಿ ರಕ್ತ ಸಂಬಂಧಿಕರೇ?	ಹೌದು/ ಇಲ್ಲ
2.	ನಿಮ್ಮ ಕುಟುಂಬದಲ್ಲಿ ಯಾರಿಗಾದರು ಶ್ರವಣ ದೋಷ, ಮಾತು ಮತ್ತು ಭಾಷಾ ಬೆಳವಣಿಗೆಯಲ್ಲಿ ನಿಧಾನಗತಿ, ಅಥವಾ ಬುದ್ದಿಮಾಂದ್ಯತೆ ತೊಂದರೆ ಇದೆಯೇ?	ಹೌದು/ ಇಲ್ಲ
3.	ಮಗುವಿನ ತಾಯಿ ಗರ್ಭಿಣಿಯಾಗಿದ್ದಾಗ ಸಮಯದಲ್ಲಿ, ಯಾವುದಾದರು ಅನಾರೋಗ್ಯದಿಂದ ನರಳಿದ್ದರೇ?	ಹೌದು/ ಇಲ್ಲ
4.	ಮಗುವಿನ ತಾಯಿ ಗರ್ಭಿಣಿಯಾಗಿದ್ದಾಗ, ಮೈಸಿನ್ ವಿಭಾಗಕ್ಕೆ ಸೇರುವ ಯಾವುದಾದರೂ ಮಾತ್ರೆ/ಔಷಧವನ್ನು ತೆಗೆದುಕೊಂಡಿದ್ದರೇ?	ಹೌದು/ ಇಲ್ಲ
5.	ಮಗುವಿನ ತಾಯಿ ಗರ್ಭಿಣಿಯಾಗಿದ್ದಾಗ ಯಾವುದಾದರು ಅನಾರೋಗ್ಯ ಇದ್ದು ಅದಕ್ಕಾಗಿ ಕಿವಿಗೆ ಹಾನಿಕಾರಕವಾಗಿರುವ ಔಷಧಿಯನ್ನು ಸೇವಿಸಿದ್ದರೇ ?	ಹೌದು/ ಇಲ್ಲ

6.	ತಾಯಿ ಗರ್ಭಿಣಿಯಾಗಿದ್ದಾಗ ಮದ್ಯಪಾನ ಅಥವಾ ಧೂಮಪಾನವನ್ನೇನದರೂ ಮಾಡಿದ್ದರೇ?	ಹೌದು/ ಇಲ್ಲ
7.	ತಾಯಿ ಗರ್ಭಿಣಿಯಾಗಿದ್ದಾಗ, ಮಾನಸಿಕ ತೊಂದರೆಗೆ ಒಳಗಾಗಿದ್ದರೇ?	ಹೌದು/ ಇಲ್ಲ
8.	ಗರ್ಭಪಾತದ ಪ್ರಯತ್ನವೇನಾದರೂ ನಡೆದಿತ್ತೇ?	ಹೌದು/ ಇಲ್ಲ
9.	ವೈದ್ಯರು ನಮೂದಿಸಿದ ದಿನಾಂಕಕ್ಕಿಂತ ಮುಂಚಿತವಾಗಿಯೇ ನಿಮ್ಮ ಮಗುವಿನ ಜನನವಾಗಿದೆಯೇ? (36 ವಾರಗಳಿಗಿಂತ ಮುಂಚೆ)?	ಹೌದು/ ಇಲ್ಲ
10.	ಮಗು ಹುಟ್ಟಿದ ತಕ್ಷಣ ಅಳಲಿಲ್ಲವೆ?	ಹೌದು/ ಇಲ್ಲ
11.	ನಿಮ್ಮ ಮಗು ಹುಟ್ಟಿದಾಗ ಕಾಮಾಲೆ ಖಾಯಿಲೆ ಬಂದಿತ್ತೆ?	ಹೌದು/ ಇಲ್ಲ
12.	11 ನೇ ಪ್ರಶ್ನೆಗೆ, ಉತ್ತರ "ಹೌದು" ಎಂದಾದಲ್ಲಿ, ಮಗುವಿಗೆ ಚಿಕಿತ್ಸೆ (ಫೋಟೊ ಥೆರಪಿ) ಮಾಡಿಸಿದ್ದೀರಾ?	ಹೌದು/ ಇಲ್ಲ
13.	ಮಗು ಹುಟ್ಟಿದಾಗ ತೂಕ ಕಡಿಮೆ ಇತ್ತೇ? (ಎರಡೂವರೆ ಕೆ.ಜಿ ಗಿಂತ ಕಡಿಮೆ)	ಹೌದು/ ಇಲ್ಲ
14.	ಮಗುವು ಹುಟ್ಟಿದಾಗ ತಲೆ, ಕಿವಿ, ತುಟಿ, ಕಣ್ಣು ಅಥವಾ ಮುಖದ ಬೇರೆ ಅಂಗಾಂಗಗಳಲ್ಲಿ ಊನತೆಗಳು ಕಂಡು ಬಂದಿದ್ದವೇ?	ಹೌದು/ ಇಲ್ಲ
15.	ಮಗುವು ಹುಟ್ಟಿದ ನಂತರ, ಚಿಕಿತ್ಸೆಗೆಂದು ಆಸ್ಪತ್ರೆಯಲ್ಲಿ ಇರಿಸಿಕೊಳ್ಳಲಾಗಿತ್ತೇ?	ಹೌದು/ ಇಲ್ಲ
16.	ಮಗುವು ಹಾಲು ಕುಡಿಯಲು ನಿರಾಕರಿಸುತ್ತಿತ್ತೆ ಅಥವಾ ಕಷ್ಟ ಪಡುತ್ತಿತ್ತೇ?	ಹೌದು/ ಇಲ್ಲ
17.	ಮಗುವು ತುಂಬಾ ಅಳುತ್ತಿತ್ತೇ ಅಥವಾ ಆಗಾಗ ಅಳುತ್ತಾ ಇರುತ್ತಿತ್ತೇ?	ಹೌದು/ ಇಲ್ಲ

1 ತಿಂಗಳಿಂದ 3 ವರ್ಷಗಳವರೆಗೆ

	ಅಪಾಯಕಾರಕ ಅಂಶಗಳು	
1.	ಮಗುವಿನ ಕುಟುಂಬದಲ್ಲಿ ಯಾರಿಗಾದರೂ ಬಾಲ್ಯದಿಂದಲೇ ಮಾತು ಹಾಗೂ ಶ್ರವಣ ಶಕ್ತಿಗೆ ಸಂಬಂಧಪಟ್ಟ ಶಾಶ್ವತವಾದ ನ್ಯೂನತೆಗಳೇನಾದರೂ ಇದೆಯೇ? (ಉದಾ: ನರದ ಕಿವುಡುತನ. ಮಾತು ಮತ್ತು ಭಾಷಾ	ಹೌದು/ ಇಲ್ಲ

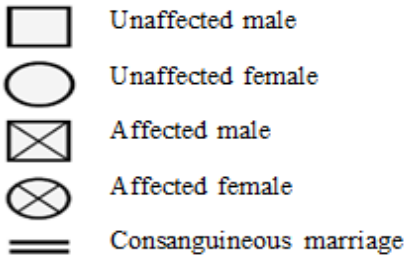
	ಬೆಳವಣಿಗೆಯಲ್ಲಿ ನಿಧಾನಗತಿ, ಬುದ್ದಿಮಾಂದ್ಯತೆ ಅಥವಾ ದೈಹಿಕ ಬೆಳವಣಿಗೆಯಲ್ಲಿ ನಿಧಾನಗತಿ ಇತ್ಯಾದಿ	
2.	ಮಗುವಿನ ತಾಯಿ ಗರ್ಭಿಣಿಯಾಗಿದ್ದಾಗ, ಯಾವುದಾದರೂ ಸೋಂಕು ತಗುಲಿತ್ತೆ?	ಹೌದು/ ಇಲ್ಲ
3.	ಮಗುವು ಹುಟ್ಟಿದಾಗ, ಅದರ ಚರ್ಮ ಹಳದಿ ಬಣ್ಣಕ್ಕೆ ತಿರುಗಿತ್ತೆ?	ಹೌದು/ ಇಲ್ಲ
4.	ಮಗುವು ಹುಟ್ಟಿದಾಗ, ತಲೆ ಅಥವಾ ಮುಖದ ಭಾಗಗಳಲ್ಲಿ ಊನತೆಗಳು ಕಂಡುಬಂದಿದ್ದವೇ?	ಹೌದು/ ಇಲ್ಲ
5.	ಮಗುವಿಗೆ ಮೆದುಳು ಜ್ವರ, ಸೀತಾಳೆ ಅಥವಾ ಸಿಡುಬಿನಂತಹ ರೋಗಗಳು ಕಂಡುಬಂದಿದ್ದವೇ?	ಹೌದು/ ಇಲ್ಲ
6.	ಮಗುವಿಗೆ ಮುಖದಲ್ಲಿ ಅಥವಾ ಕಿವಿಯ ನಾಳದಲ್ಲಿ ಊನತೆಗಳು ಕಂಡುಬಂದಿದ್ದವೇ?	ಹೌದು/ ಇಲ್ಲ
7.	6ನೇ ಪ್ರಶ್ನೆಗೆ ಉತ್ತರ ಹೌದು ಎಂದಾದಲ್ಲಿ, ಮಗುವಿಗೆ ಶಸ್ತ್ರ ಚಿಕಿತ್ಸೆಯೇನಾದರೂ ಮಾಡಿಸಿದ್ದೀರಾ?	ಹೌದು/ ಇಲ್ಲ
8.	ನಿಮಗೇನಾದರು ಮಗುವಿನ ಮಾತು ಮತ್ತು ಭಾಷಾ ಬೆಳವಣಿ ವಯಸ್ಸಿಗೆ ತಕ್ಕಂತೆ ಇಲ್ಲ ಎಂದು ಅನ್ನಿಸಿದ್ದು ಇದೆಯೇ?	ಹೌದು/ ಇಲ್ಲ
9.	ಮಗುವು ಆದೇಶಗಳನ್ನು ಪಾಲಿಸಲು ಕಷ್ಟ ಪಡುತ್ತದೆಯೇ?	ಹೌದು/ ಇಲ್ಲ
10.	ಮಗುವಿನ ಶ್ರವಣ ಶಕ್ತಿ, ಮಾತು ಮತ್ತು ಬೇರೆ ಬೆಳವಣಿಗೆಗಳ ಬಗ್ಗೆ ತಂದೆ ತಾಯಿ/ಪೋಷಕರಲ್ಲಿ ಏನಾದರು ಅನುಮಾನವಿದೆಯೇ?	ಹೌದು/ ಇಲ್ಲ
11.	ಮಗುವಿನ ಮಾತು ಮತ್ತು ಭಾಷೆಯ ಬೆಳವಣಿಗೆಗೆ, ಮನೆಯವರಿಂದ ಕಡಿಮೆ ಅಥವಾ ಸಮಂಜಸವಲ್ಲದ ಪ್ರೋತ್ಸಾಹ ದೊರೆಯುತ್ತಿದೆಯೇ?	ಹೌದು/ ಇಲ್ಲ
12.	ಮಗುವಿಅ ತಲೆಗೆ ಪ್ಟ್ಟು ಬಿದ್ದು ಜ್ಞಾನ ತಪ್ಪಿದ್ದು/ತಲೆಬುರುಡೆ ಅಪಘಾತವಾಗಿ, ಅದರಿಂದ ಕಿವಿತಲ್ಲಿ ರಕ್ತ ಸೋರಿದ್ದು ಉಂಟೇ?	ಹೌದು/ ಇಲ್ಲ
13.	ಮಗುವಿಗೆ ಸೆಳೆವು ಏನಾದರೂ ಬಂದಿತ್ತೇ?	ಹೌದು/ ಇಲ್ಲ
14.	ಮಗುವಿಗೆ ಮೂರು ತಿಂಗಳುಗಳ ಕಾಲ ಕಿವಿ ಸೋರಿದ್ದು ಉಂಟೇ?	ಹೌದು/ ಇಲ್ಲ
15.	ಮಗುವು ಆಟಿಕೆಗಳನ್ನು ತಲುಪಲು ಅಥವಾ ಸರಿಯಾಗಿ ಉಪಯೋಗಿಸಲು ಕಷ್ಟಪಡುತ್ತದೆಯೇ? (ಉದಾ: ಆಟಿಕೆಗಳನ್ನು ಜೋಡಿಸುವುದು?)	ಹೌದು/ ಇಲ್ಲ

16.	ಮಗುವಿಗೆ ಅಗಿಯಲು, ಕುಡಿಯಲು ಮತ್ತು ನುಂಗಲು ತೊಂದರೆ ಉಂಟಾಗುತ್ತದೆಯೇ?	ಹೌದು/ ಇಲ್ಲ
17.	ನಿಮ್ಮ ಮಗುವು ಅತಿ ಚಟುವಟಿಕೆಯಿಂದ ಇರುತ್ತದೆಯೇ?	ಹೌದು/ ಇಲ್ಲ
18.	ಮಗುವಿಗೆ ಪದಾರ್ಥಗಳು, ಮನುಷ್ಯರು, ಶಬ್ದ, ರುಚಿ ಅಥವಾ ವಾಸನೆಗೆ ಅಸಹಜವಾದ ಆಕರ್ಷಣೆ ಇದೆಯೇ?	ಹೌದು/ ಇಲ್ಲ
19.	ಮಗುವಿನ ನಡುವಳಿಕೆಯಲ್ಲಿ ತೊಂದರೆ ಏನಾದರೂ ಇದೆಯೇ?	ಹೌದು/ ಇಲ್ಲ
20	ಮಗುವು 2-3 ವರ್ಷವಿದ್ದಾಗ, ಉಗ್ಗುವಿಕೆ ಕಂಡುಬಂದಿತ್ತೆ?	ಹೌದು/ ಇಲ್ಲ

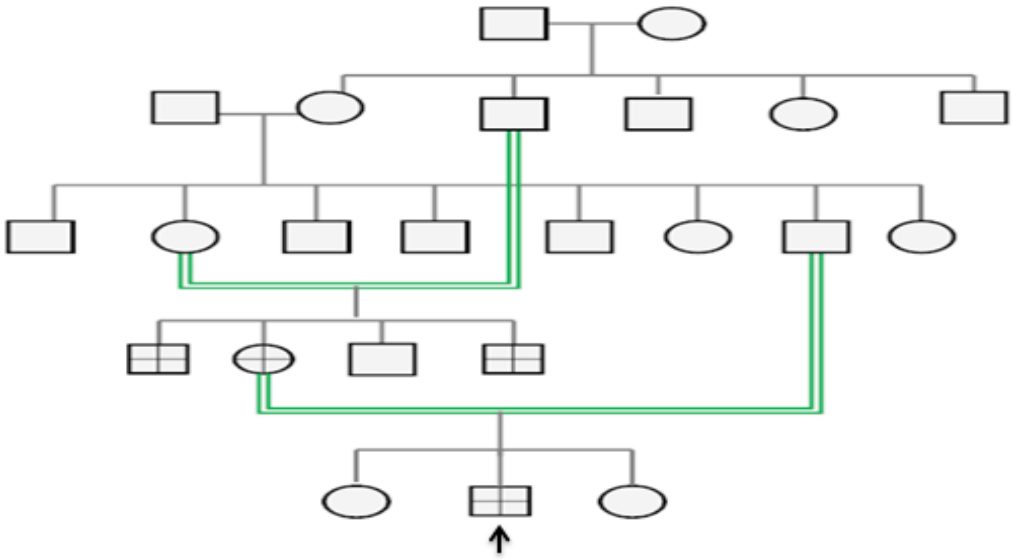
Appendix C

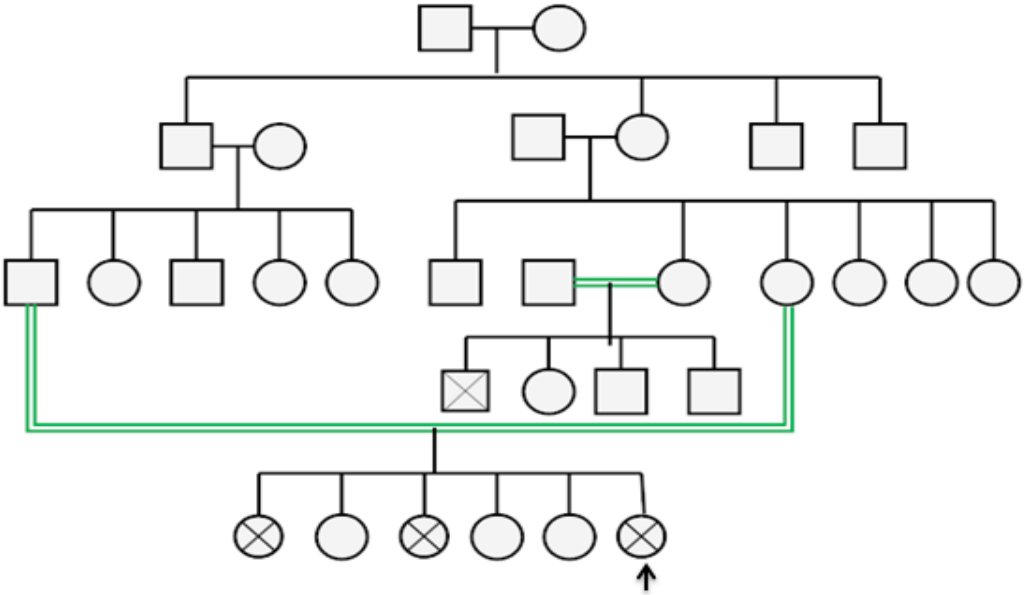
Examples of Pedigrees of cases with Autosomal Recessive, Autosomal dominant, Cytoplasmic and Holandric type of gene inheritance .

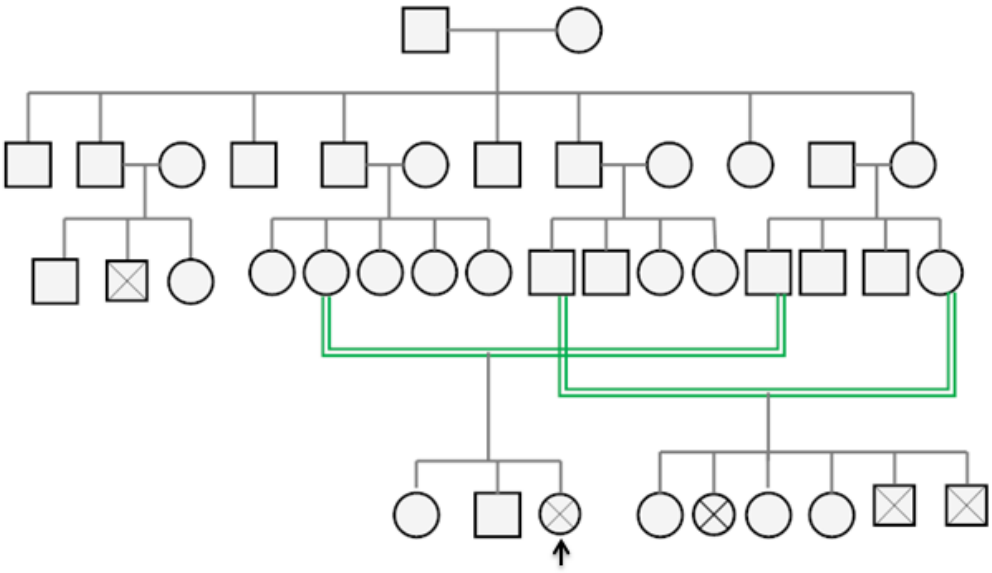
Note

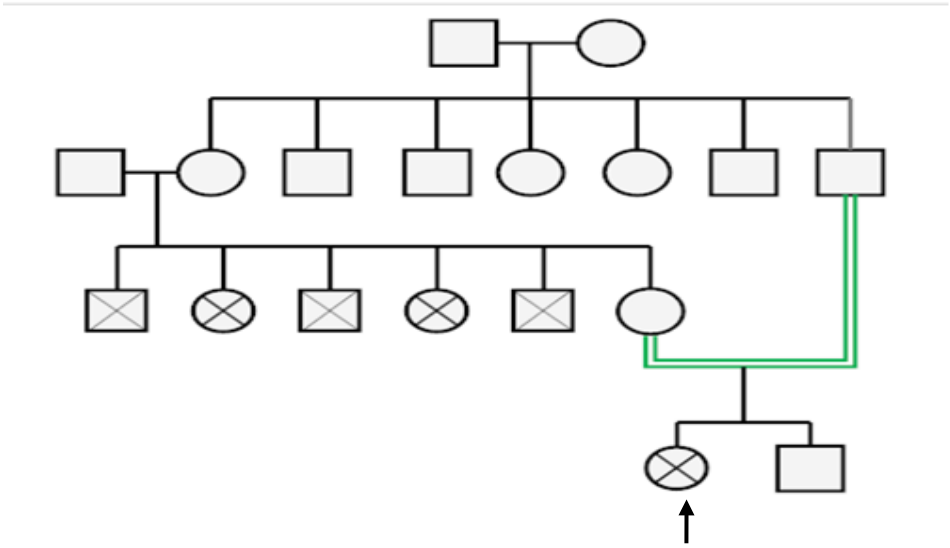


Pegrees of cases with autosomal recessive type of inheritance .

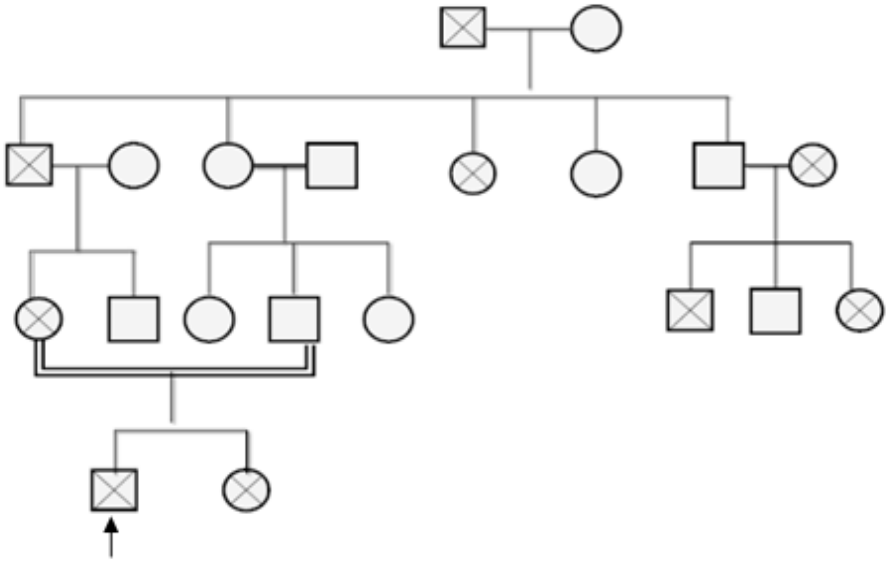


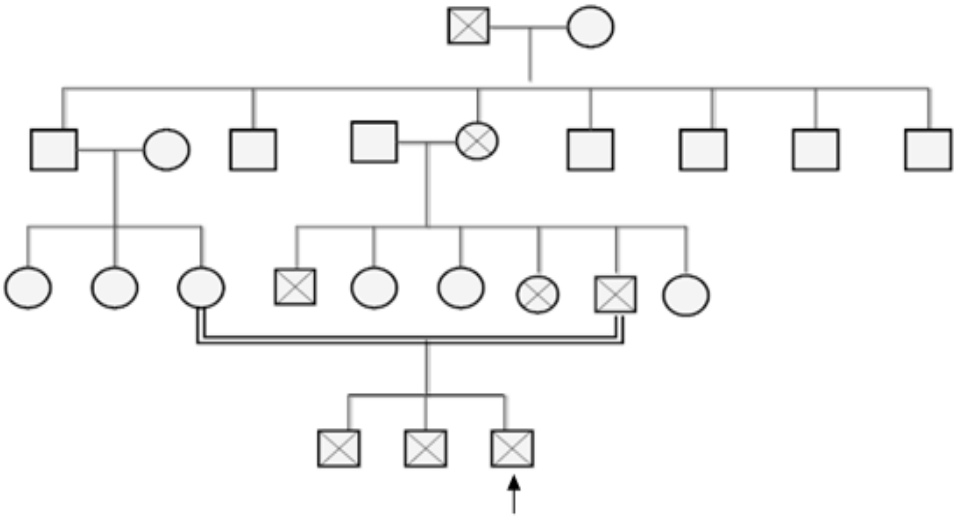




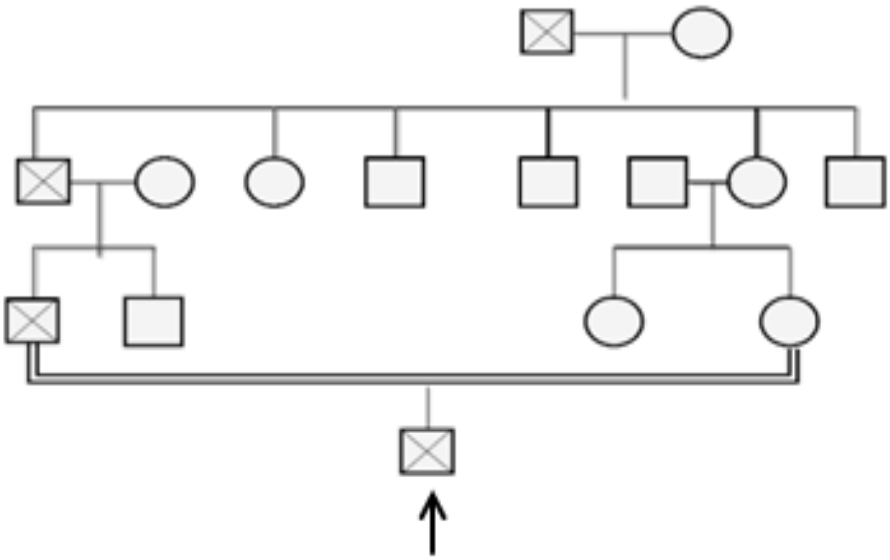


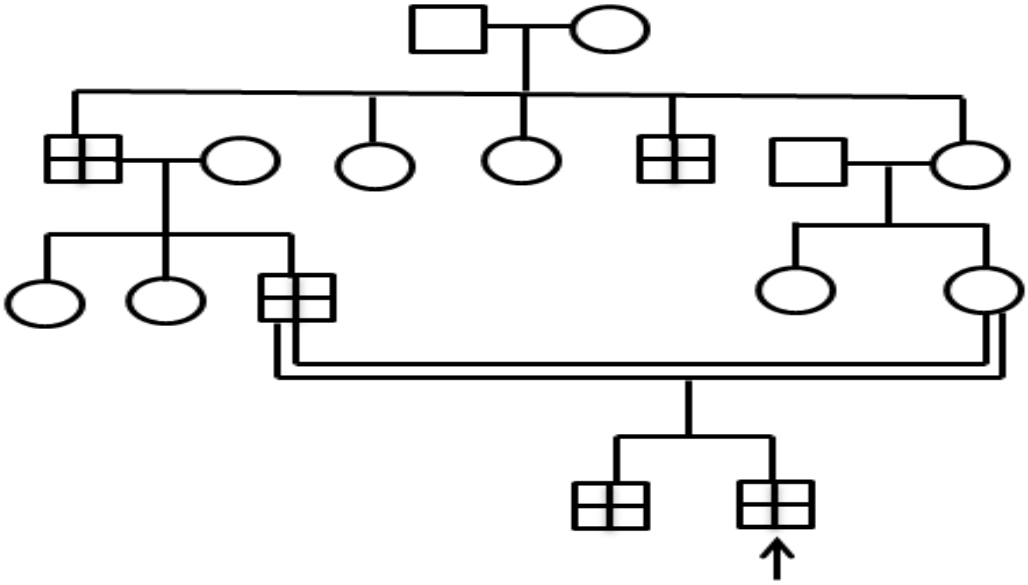
Pegree of cases with **autosomal** dominant type of inheritance.



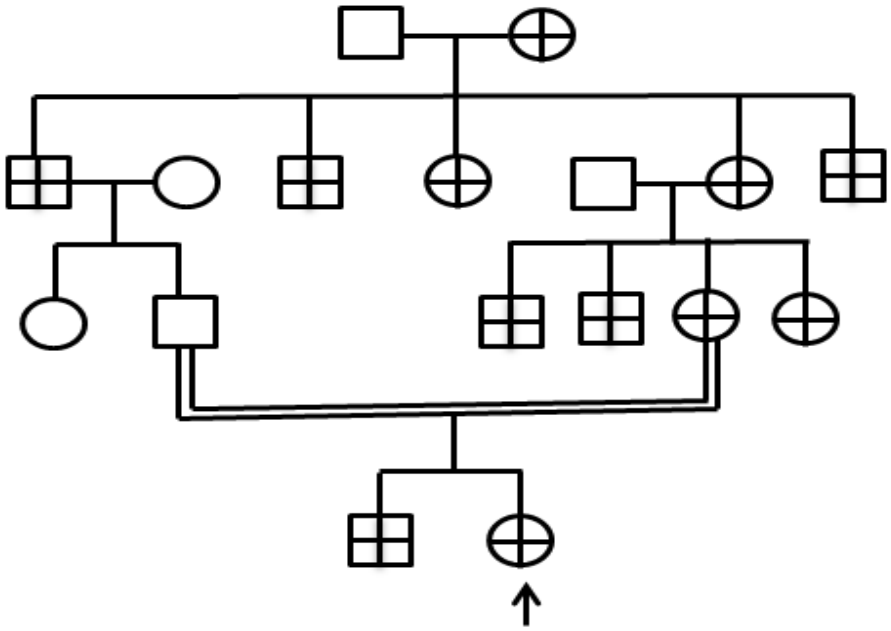


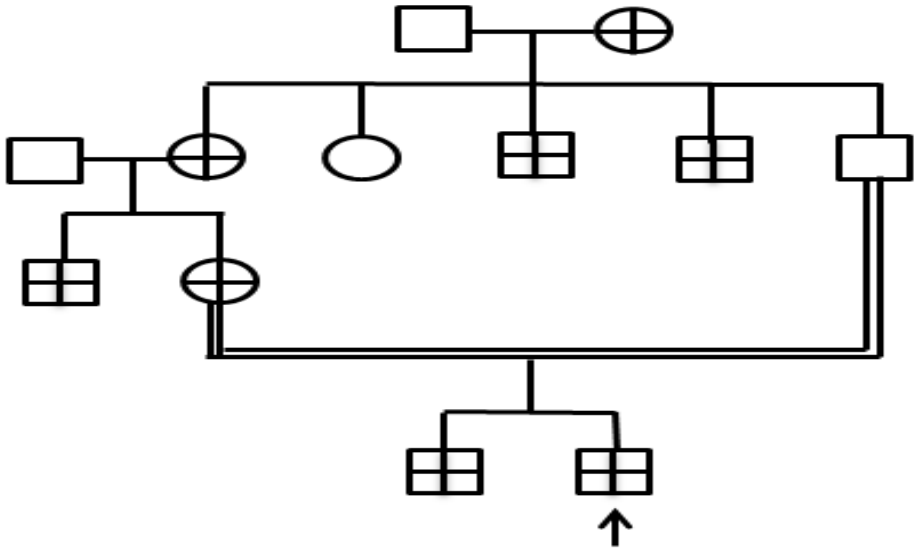
Pegree of cases with Holandric (Y-linked) type of inheretence.





Pegree of cases with Cytoplasmic (X-linked) type of Inheritance





Appendix –D

List of communication disorders included in the study

Sl No	Communication disorders
1.	DSL secondary to HL: Delayed speech and language secondary to hearing impairment
2.	DSL secondary toID: Delayed speech and language secondary to intellectual disability
3.	SLI: Specific language impairment
4.	DSLsecondary to CP: Delayed speech and language secondary to cerebral palsy
5.	SSD: Speech sound disorder
6.	DSL secondary toASD: Delayed speech and language secondary to Autism spectrum

	disorder
7.	DSLsecondary toSD: Delayed speech and language secondary to seizure disorder
8.	FD: Fluency disorder
9.	LD: Learning disability
10.	DSL secondary to CLP: Delayed speech and language secondary to Cleft lip and palate
11.	DSL secondary toSynd: Delayed speech and language secondary to syndrome.