

Impact of a Genetic Counseling Replica on Parents of Children with Down Syndrome: A South Indian Cohort Study

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ABSTRACT Parents having Down syndrome children may not be aware of the reason for their child's present condition and knowledge to hold the situation hence, in such conditions genetic counseling is important to guide the parents about caring for the child with Down syndrome. So the main aim of the researchers' study is to understand the impact of Genetic counseling replica on the parents having Down syndrome in a language they understand better and also to understand the parent's experience of Genetic counseling and knowledge of Down syndrome. The Genetic counseling session was carried out in two phases attended by eight parents having Down syndrome children and after Genetic counseling session parents were able to understand possible condition that may be the reason for their child's current situation. The researchers' study concluded that Genetic counseling should be the best replica to help the parents having children with Down syndrome.

INTRODUCTION

According to the GC Definition Task Force of the National Society of Genetic Counselors, this process integrates the following: interpretation of family and medical histories to assess the chance of disease occurrence or recurrence, education about inheritance, testing, management, prevention, resources and research, and the promotion of informed choices and adaptation to the risk or condition (Resta et al. 2006). It is known that ethnic identity may influence perceived benefits and barriers related to genetic testing and GC. Thus, GC services should take these factors into account and create culturally-appropriate conditions which best meet the needs of heterogeneous patient populations (Alsulaiman et al. 2012). Down's syndrome (DS) is the most common postnatal viable human autosomal chromosomal abnormality with an estimated 70–80 percent prenatal lethality (Galdzicki and Siarey 2003). There are various conserved

features occurring in all DS population, including learning disabilities, craniofacial abnormality and hypotonia in early infancy (Antonarakis et al. 2004). In 1959, Lejeune and colleagues discovered the genetic basis of DS named as trisomy of chromosome 21, which is the smallest human autosomal chromosome (Neri and Opitz 2009). The development of secondary sexual characteristics in DS is similar to other adolescents. The fetal oogenesis of women with the syndrome appears to be normal and, therefore, they are capable of reproduction (Jagiello et al. 1987). On the other hand, men have diminished reproductive capacity, showing testicular histology compatible with oligospermia and, frequently, hypogonadism (Mercer et al. 2004). Genetic counseling is also important to guide the parents about caring for the child with DS. Because individuals with DS often experience delays in reaching various developmental milestones, early intervention with speech therapy, occupational therapy, and physical therapy is recommended as it maximizes long-term outcomes (Rihtman et al. 2010). It is recommended that families of children with Down syndrome (DS), intellectual disability (a term used to refer to those with mental retardation [MR]), or autism spectrum disorder (ASD) be offered a comprehensive medical genetics evaluation that includes genetic counseling (Simonoff 1998) and genetic testing (Filipek et al. 2000). The effectiveness of

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the GC process depends not only on the social and cultural aspects but on the professional's human experience and counseling abilities (Battista et al. 2011). Genetic counselors need to be aware of the factors that result in both positive and negative genetic counseling experiences. Each individual is unique in how they perceive genetic counseling, what emotional and educational needs they bring to a consultation, as well as when they require such sessions (Morris et al. 2015).

Objectives

The main aim of the researchers' study was to understand the impact of Genetic counseling replica on the parents having Down syndrome in a language they understand better and also to understand the parent's experience of Genetic counseling and knowledge of Down syndrome.

METHODOLOGY

The study was carried out at a special school having nearly 52 special children with intellectual disability and out of those children 8 are Down syndrome with intellectual disability. So then it was decided to provide Genetic counsel-

ing to the parents of those 8 special children. In the study parents were informed previously about Genetic Counseling (GC) session by the school management and the parents can ask about the necessary details regarding the current condition of their respective Down syndrome children and other necessary details. The study was approved by Institutional Human Ethical Committee of VIT University, Vellore and before starting the GC session each parents were informed about importance of GC and a written consent was obtained. The session attended by all 8 parents were from middle class family of south Indian origin and most of the parents had education upto school. The mean age of mother was 29.88, father age was 32.75, and mean age of children was 7.25. It was also found that most of the mothers are housewives and fathers are daily laborers which is given in Table 2.

The GC Session was Carried Out in Two Phases

First Phase

In this phase, one to one interview was conducted with each parents and during which following information was asked from each par-

Table 1: Family and natal history of Down syndrome children

Patients code	Type of family	Consang- uinity	Family history of distress	Term	Labour duration	Delivery type	Birth weight	Prolapsed cord	Respiratory
Case1	Nuclear	No	No	Full	Normal	Normal	Normal	No	No
Case 2	Nuclear	No	No	Full	Normal	Caesarean	Low (1.6kgs)	No	No
Case 3	Joint	Yes	No	Premature	Normal	Caesarean	Normal (2.3kgs)	No	No
Case 4	Joint	No	No	Premature	Prolonged	Caesarean	Low	Yes	No
Case 5	Joint	No	No	Post-mature	Normal	Normal	Normal	No	No
Case 6	Nuclear	Yes	No	Full	Normal	Normal	Normal	Yes	Yes
Case 7	Nuclear	No	No	Full	Normal	Normal	Normal	Yes	No
Case 8	Joint	Yes	No	Full	Normal	Normal	Normal	Yes	Yes

Table 2: Personal details of parents having Down syndrome children

	Age			Level of education		Nature of work	
	Mother	Father	Child	Father	Mother	Father	Mother
Case 1	22	27	3	School	School	Daily labor	Housewife
Case 2	25	30	5	School	School	Daily labor	Housewife
Case 3	27	33	9	School	School	Daily labor	Housewife
Case 4	21	27	2	School	School	Business	Housewife
Case 5	38	36	7	Diploma	School	Factory worker	Household work
Case 6	30	35	9	School	School	Daily labor	Tailor
Case 7	23	29	3	School	School	Auto diver	Housewife
Case 8	53	45	20	School	School	Daily labor	Housewife

ents (family history of DS, Consanguinity, type of family, pregnancy term, labor duration, delivery type, birth weight, nature of work and age of parents). See Table 1.

Second Phase

In the second phase, all the queries of parents regarding the current situation of their children were discussed and parents were asked about the satisfaction they had from Genetic Counseling session.

RESULTS

Some Highlights of Parents Opening Up during First GC Session

Parents 1: *Since 8 years we don't had children and based on the doctor advice we had couple of IVF by spending nearly lakhs rupees and after deliver we found our baby is normal but different from other baby and we didn't understand what had happened (Given in Fig.1).*

Parents 2: *After our first child condition our family is in greater shock and now we have fear to have second child.*

Parents 3: *My girl is normal in appearance from outer even she is down syndrome so we fear about their personal safety as she can be misused....so it is possible to remove her uterus.*

Parents 7: *Our family is typical joined Muslim family, we both are cousins and we found that in our family there 9 people having Intellectual disability and among which is 1 our kid. We didn't want this again to be happening in our family (Given in Fig.2).*

Parents 8: *We can't attend the social function or any other kind of family function as we have to keep our child always with us and also to make sure that others don't tease our kid.*

Parents 5: *We belong to lower middle class family; our whole thinking is about the future of our effected child and medical expense.*

Some Highlights of Satisfaction Regarding GC Given to Parents.

Parents 1: *We had a fear and wrong opinion about the IVF technique but the GC session provided better clarification on IVF and GC has also provided information regarding what had really happened during our first experience with IVF.*

Parents 3: *Genetic counseling has made us realize the importance of reproductive organ of Girl child and various complications that might have risen in case if we remove it.*

Parents 7: *The session was conducted in such a manner that we really understood about the current condition of our child and GC session also helped us by providing various information on genetics testing and prenatal testing available now.*

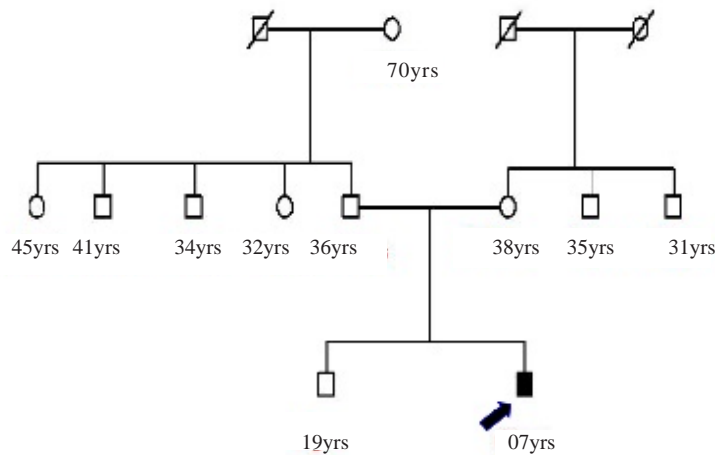


Fig. 1. Pedigree showing no consanguinity between parents having Down syndrome child

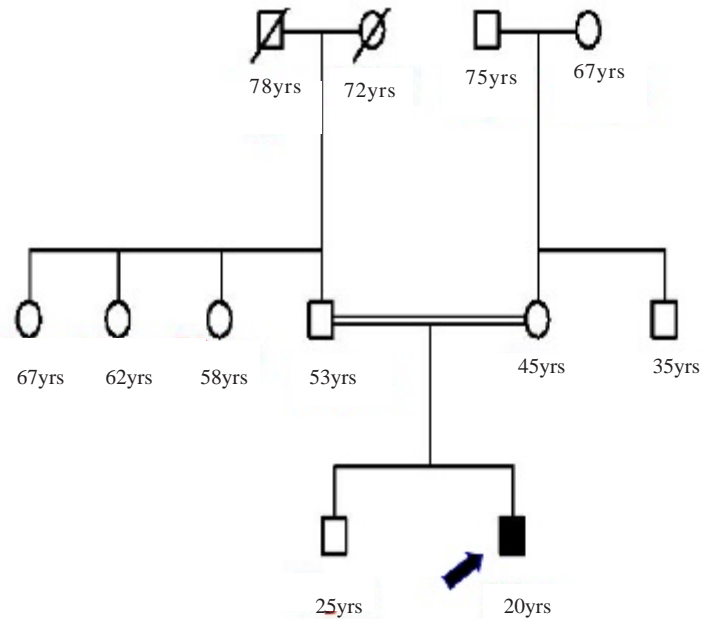


Fig. 2. Pedigree showing consanguinity between parents having Down syndrome child.

DISCUSSION

The main objective the researchers' study was to provide a Genetic counseling as a replica to the parents having children with Down syndrome with intellectual disability by providing the necessary information about the current situation of their special child in a way that parents understand better and also to get the feedback from the parents about the Genetic counseling session. In this study there were 8 parents belonging to the middle class family of south Indian origin. During this study it was found that parents were really interested to attend the session as they were having lack of knowledge on the current situation of their child and by looking at the parents it could be felt the amount of stress they were undergoing daily. One of the hallmarks of DS is the variability in the way that the condition affects people with DS. With the third 21st chromosome existing in every cell, it is not surprising to find that every system in the body is affected in some way. However, not every child with DS has the same problems or associated conditions. Parents of children with DS should be aware of these possible conditions

so that they can be diagnosed and treated quickly and appropriately (Asim et al. 2015). During the first phase of Genetic counseling session the researchers found that all the parents were much open, they said all the things which they were looking for the answer for long time and they could understand that most of parents are worried about the future of their special child, parents are also concerned about the girl child safety as anyone can mishandle and moreover almost all of them are looking for all kind of help from government to manage their special children as they all belong to lower middle class family. Parents also expressed their sadness when they heard that their born child is abnormal, this surely had shocked them as they had lot of future plans for their child. As healthcare has improved for individuals with DS, the average life expectancy has increased by more than 30 years, from an average of 25 years of age in 1983 to almost 60 years of age in 2000 (Rihtman et al. 2010). During the Genetic counseling session the researchers found that parents were not aware of the current medical treatment or therapies to treat their kids and it's their study which has provided all the necessary things

which parents can do to give quality life to their children. Genetic counselors should balance the negative aspects of DS, such as birth defects, medical complications, and developmental delay, with positive aspects like available treatments, therapies, and the ability for people with DS and their families to enjoy a high quality of life (Bryant et al. 2001). The researchers' study also recommends adopting genetic counseling as a replica to help the parents having children with birth defect or intellectual disability like Down syndrome for proper counseling of parents to overcome the pressure handling the current situation of their child. Counseling for parents in the newborn period is well covered in the commentaries by the McCabe and McCabe and Van Riper and Choi. To reiterate, it is important to congratulate the parents for having a new baby and assure them that the doctor/ genetic counselor are there to support them (Van Riper et al. 2011; McCabe et al. 2011). As suggested by Van Riper and Choi some discussion regarding the variability in adaptation of parents in raising a special-needs child needs to occur (Van Riper and Choi 2011). The researchers' study also found that some of parents had consanguinity marriage; among those was one couple who were first degree cousins and they found that in their family 9 people had intellectual disability and main reason could be consanguinity marriage and so it was suggested to them that Genetic counseling could help their family better. Consanguineous marriages that are rare in developed countries are still a GC challenge in some populations with around 1.1 billion people currently living in countries where consanguineous marriages are customary (Strauss 2009). The researchers' study also found that in India we don't have proper guidelines for any health care expert to handle the situation like Down syndrome or other birth defects. The National Society of Genetic Counselors recently developed a list of guidelines that healthcare providers should follow when delivering a diagnosis of Down syndrome (Sheets et al. 2011). The guidelines follow many of the tenets of appropriate Genetic counseling including the central tenet of non-directiveness of the counselor, which requires the counselor to be neutral and balanced in the presentation of the information in order to support and respect the patient's values and decisions (Sheets et al. 2011). These guidelines include the following: 1.) Tell the par-

ents about the diagnosis as soon as possible, 2.) Deliver the diagnosis in person by a healthcare provider with sufficient knowledge about the condition, 3.) Meet with both parents together, or arrange a telephone call at a time when both partners will be present, 4.) Inform the family of the diagnosis in their preferred language, 5.) Discuss the diagnosis in a private, comfortable setting, 6.) Provide parents with accurate and up-to-date information, 7.) Provide the information in a sensitive and caring, yet confident and straightforward manner, 8.) Use neutral language and avoid value judgments, 9.) Use sensitive language and avoid outdated or offensive language, 10.) Allow time for silence and tears, 11.) Assess the emotional reaction of the parents, and validate those feelings, and 12.) Provide informational resources [18]. The South African Human Genetic Policy Guidelines include a rudimentary list pertaining to the management of DS (Antonarakis et al. 2004). In the USA, the American Academy of Pediatrics (AAP) has addressed this issue by devising clinical protocols focusing on improving the level of functioning and quality of life of children with DS (Cunniff et al. 2001). The study by Morris et al. (2015) on the mother's experiences of genetic counseling in Johannesburg, South Africa concluded that greater effort is needed to create public awareness of genetic services and genetic conditions in South Africa. The information provided to individuals during genetic consultations should be locally and personally relevant, and on-going patient support may be required, particularly when explaining the condition to family and community members.

CONCLUSION

The researchers' study concluded that Genetic counseling should be the best replica to help the parents having children with Down syndrome as parents were very satisfied with the Genetic counseling because it was given in the best language each parent understands.

RECOMMENDATIONS

Recommendations for future research include using large sample size with different ethnic background for investigation of the experiences of individuals seen for genetic counseling for other genetic conditions in various other parts of

India. Genetic counseling should be given to the people according to the language they understand better and also according to the knowledge of each individual.

LIMITATIONS OF THE STUDY

The main limitation of the current study was small sample size, as the study comprised of only eight parents having Down syndrome children and also from same cohort.

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