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ORIGINAL ARTICLE

Knowledge and Awareness Regarding Trisomy 21 among Parents of School Going Down Syndromes in Islamabad and Rawalpindi: a Descriptive Cross-Sectional Study

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ABSTRACT

Background: Down syndrome or trisomy 21, is mostly associated with characteristic facial features, delayed physical growth and mild to moderate intellectual disability with co-morbidities. Not many people are well aware of facts related to down syndromes.
Aim: To identify the level of awareness and knowledge among parents of school going down syndrome.
Methodology: A descriptive cross-sectional survey conducted at special children's schools in Islamabad and Rawalpindi, from February 2021 to March 2021. Sample size was 44. We adopted convenience non-probability sampling technique. Parents that fulfill the inclusion criteria i.e. school going down syndromes, 5 to 32 years of age and residents of Islamabad and Rawalpindi, in an interactive counselling session, keeping the questionnaire anonymous. After data collection it was entered in SPSS v25 for statistical analysis.
Results: Analysis of the questionnaire regarding the Knowledge of physical features of a child with Down syndrome showed that most 97% of the people were aware about facial profile and nose. 65.9% people responded positively regarding their knowledge about small head, ears and nose. 88.6% had knowledge about wide, short hands and short fingers. 65.9 knew about single deep crease across palm. 88.6 told that Down's children had long tongue. 45.5% knew they have squint. 88.6% have knowledge about loss of muscle tone. Regarding memory 45.5% knew they have poor memory while 93.2% knew they are slow learners.
Conclusion: Our selected candidates are mostly well aware of this condition but there is still the need for spreading further awareness to overcome the problems faced by the child and for their better management.
Keywords: Awareness, Down Syndrome, Communication

INTRODUCTION

Down Syndrome (DS) is one of the foremost mental retardations that occur genetically at the time of birth leading to intellectual deformity. The earliest reported incidence of people with DS was in 1838 which described the intellectual and developmental disability and referred them as idiots as being separate from insanity[1]. However, it was in 1866 when the English physician Dr. John L. H. Down described detailed physical characteristics of people with DS. John Down identified the DS patients as ethnic Mongols because of their resemblance to Mongolian people, therefore the term mongolism or mongoloids was used in the academic research[2]. In 1959, a French physician Dr. Jerome Lejeune discovered the landmark 'trisomy 21'; which is the presence of an extra chromosome-21 in the DS patients[3]. Finally, in 1961, it was termed 'Down's syndrome or anomaly', or 'trisomy 21 anomaly'[4]. An article by Antonarakis et al. provides a detailed overview of different genotypes that cause anomalies in patients with the DS which result differ in different clinical manifestations of the DS. Moreover, it also contributes towards the health complications and comorbidities of other defects at the time of birth of people with DS[5].

The incidence in general population of having a child with DS is one in 1,000[5]. However, it varies according to the geographical distribution as well. Incidence is one in 732 infants in the US born with DS[6]. One in 865 in Nigeria, one in 500 in South Africa, one in 319 in United Arab Emirates, one in 500 in Oman, one in 554 in Saudi Arabia, one in 500 in China, one in 3000 in Hong Kong, one in 1200 in Mumbai, 0.92-1.24 in 1000 in Pakistan, one in 650 in Mexico, 1.41 in 1000 in Canada, 1 in 405 in Chile, and one in 735 in Uruguay[7]. It is yet unknown the reason for the variance in geographical regions. However, the highest rates of incidence found in Arabs, primarily hypothesized by Alosaimi et al. is due to the consanguineous marriages, secondly due to refusal for prenatal testing. They also found multiparity to be a major contributing factor towards the incidence of DS in Saudi Arabia. To the contrary, Europe has the lowest rates of occurrence, primarily because prenatal screening and acceptance of abortion in cases pertaining to DS[8]. It is widely accepted amongst all the researchers that maternal age plays the foremost role in the probability of conceiving fetus with DS characteristics. The incidence varies from having one in 2000 at the age of 20 to one in 10 at an age of 49[9]. Al-Bilgati debated the age of maternal grandmother that can disturb the meiosis of the daughter due to the disturbances in grandmother's gonads, which results in trisomy 21 in daughter's oocytes. Similarly, father' increasing age can be an additional factor, which somehow is compensated by younger age of the mother. Other acquired factors are smoking, use of drugs, unavailability of prenatal screening and antenatal screening[7].

In a survey conducted by Skoto out of >1,000 mother respondents, 87% reported that they were informed about their child having DS after the delivery. Hardly anyone expressed the feeling of joy, to the contrary, these mothers were frightened about the lives of their child. Over the period of time, and with the support of family, siblings, and other mothers of DS patients, the fear diminishes and turns into the support for their child[10]. Alosaimi et al. found that 4 out of 5 mothers in Saudi Arabia does not know that children with DS requires special care, and 3 out of 5 mothers did not know the causality of DS. Alosaimi et also discussed that in Saudi Arabia majority of parents were informed with the diagnosis of their child with DS after the delivery by pediatrician, obstetric or gynecologist[8]. The same complain is noted in the study by Skoto and Hall S that parents were informed about the diagnosis of child either after birth or complains of developmental delay and one in two mother got depressed after finding out that their child has DS respectively[11].

John and Gayatri found in their survey in India that 95% of the family believe that their child with DS can be sent to regular school with normal kids, while 90% believe that their child with DS can be trained, and 98% responded that these kids can be trained[12]. Haider and Khan found that parental motivation has a favorable impact on the development of the child with DS, while the parent's education level itself matters as compared to an uneducated parent[13]. Ruiz-González Et Al. (2019) discussed that high body mass index (bmi) and obesity is an evident issue in across all the people with DS, therefore, awareness of some form of physical activity is essentially required[14]. Silva et al. discussed that motor development in children with DS is less than the children without the DS[15].

Alosaimi et al. (2020) discussed that societal response towards people with DS is unfavorable, mainly due to lack of awareness and knowledge about the congenital defect[8]. Hall S found in their study that family of the child with the DS has blamed the mother for the congenital defect in the child[15]. Urbano and Hodapp (2007) in a comparative study among families having a child with DS, other birth defects and no identified disability found that divorce rates to be lesser as compared to the other two groups, however, age, education and demographics plays a contributing role. This is known as 'Down-syndrome advantage'[16]. Burke et al. analyzed the Tennessee birth records to deduce that family with their first child born with DS were 45.7% likely to have a subsequent child[17]. Participants in a study conducted by Levis et al. (2012) expressed their fear of having such child, as it will disrupt the quality of their lives by increasing the stress and financial burden. While the same participants expressed that having a child with DS would be beneficial for both normal children as it will teach them empathy, while child with DS will learn to communicate in a diverse situation[18].

People with DS manifests over fifty distinctive features that varies child to child, however most of the people does have very fundamental knowledge about it, even in an advanced and developed country like the US.Children with DS are amiable, pleasant, and social; however, they do go with the same level of emotional development, therefore, selective conditioning is required with them. Duranovic et al. mentions that children with DS are less creative, therefore, the role of an educator plays an integrative role by indulging them into interactive games and exercise[19].

METHODOLOGY

A descriptive cross-sectional study was carried out at special children's schools in Islamabad and Rawalpindi, from February 2021 to March 2021. It was designed after systemic literature review. The systemic literature review was done using online databases such as pub-med, google scholar, and the library of Al-Nafees Medical College. Considering a limited number of facilities, we adopted convenience non-probability sampling technique to study population. Parents of these children who fulfilled the inclusion criteria were interviewed. Parents of 44 school going Down's syndromes children and residents of Islamabad and Rawalpindi were included, in an interactive session. The included age group of the students with DS, was from 5 to 32 years of age, their diagnosis was reconfirmed through school documents, along with confirmation by the doctor interviewing their parents. An awareness workshop was conducted to assess the awareness and knowledge regarding trisomy 21 among parents of school going down syndromes at Special Children Bahria College Islamabad and Sedum Special Children School Rawalpindi. The workshop was an interactive counseling session. The authors conducted the counseling session in collaboration with the pediatrician. Informed verbal and written consent was taken prior to the session from the institutes and the parents. The questionnaire was kept anonymous.

The awareness workshop consisted of two sessions. During the first session, a survey questionnaire was handed over to the participants. The questionnaire helped us to gather the information that was required for our descriptive cross-sectional study. The questionnaire's first part was based on demographic questions. In contrast, the second part of the questionnaire consisted of series of questions assessing the knowledge and awareness regarding down syndromes. The second part of the session focused on giving information and helping parents understand what DS is and how to manage their DS children in different daily life scenarios.

After data collection, it was entered in SPSS v25 for statistical analysis to document frequencies and percentages of study variables.

RESULTS

The study enrolls 44 participants who answered the questionnaire with the informed consent. The statistical analysis revealed that amongst parents 23(52.3%) were mothers of child with DS, while 21(47.7%) fathers answered the knowledge and awareness questionnaire. It is substantial to evaluate the educational status of the DS parents to assess the knowledge regarding their child. Primary education was completed by 4(9.1%) participants, while 22(50%) parents achieved secondary education, 12(27.3%) were graduate and only 6(13.6%) parents hold post-graduation degree. The demographic results include the mother's age range between 20 years to 41 years, mean (SD) 26.5 (5.0), socio economic background of the participants was mainly lower middle class 20(45.5%) while 17(38.6%) belongs to upper middle class, and only 7(15.9%) participants belong to elite class. The mean (SD) age of their DS child is 15.72 (7.6) ranging between 5 years to 32 years. with average of mean (SD) weight 40.42 Kg (19.2) ranged from 16-89 Kgs. mean height (SD) of 53.56 inches (6.69) ranged from 32-67 Inches.

Regarding the knowledge assessment of the physical characteristics, Figure 1 demonstrates the details of knowledge of parents in context to flattened face, small head, wide short hands, single deep crease across the palm, long tongue, squint, loss of muscle tone, poor memory and slow learning capacity associated with DS. It explicit the awareness of possibilities of physical characteristic compared with the presence of specific characters in their child.

Regarding awareness of this disease 44(100%) parents believed that DS is genetically transmittible from parents to offspring. The statistics revealed that 44(100%) parents acknowledge that DS is manageable. Furthermore, 40(90%) parents answered "NO" to possibility of pre-natal screening, while only 9.1% (n = 4) parents were aware that pre-natal screening can diagnose DS before the birth of the child. The parents perhaps lack knowledge of screening and diagnosis of the genetic phenomena. Surprisingly, 42(95.5%) parents were not informed by their doctors that the child is suspected or confirmed case of DS. Moreover, 44(100%) parents were fully aware of the DS characteristics and phenotype. Table: 2 further elaborates awareness questionnaire.

Table 1: Demographic data Relation

Data	Frequency	%age
Mother	23	52.3
Father	21	47.7

Education

Data	Frequency	%age
Primary education	4	9.1
Secondary education	22	50.0
graduate	12	27.3
postgraduate	6	13.6

Socio

Data	Frequency	%age
Low middle class	20	45.5
Upper middle class	17	38.6
Elite class	7	15.9

Age of mother

Data	Frequency	%age
20-29 years	31	70.5
30-41 years	13	29.5
Total	44	100.0

Demonstration of awareness questions were further divided into agreement, neutral response and disagreement in Table: 4.

Around 25(56.8%) parents agreed that the child born with DS are weak at birth while 19(43.2%) disagree with the statement. Majority of parents 32(72.7%) disagree that life expectancy can be improved in DS patients with improvement of the quality of life.

Baseline characteristics of child Child age

Data	Frequency	%age
5-10	12	27.3
11-20	23	52.3
21-32	9	20.5
Total	44	100.0

Gender

Data	Frequency	%age
female	21	47.7
Total	44	100.0

Weight

Data	Frequency	%age
16-26 kg	13	29.5
27-37 kg	11	25.0
38-48 kg	7	15.9
49-59 kg	4	9.1
60-70 kg	5	11.4
81-89 kg	4	9.1

Height

Data	Frequency	%age
38-48 Inches	12	27.3
49-59 Inches	25	56.8
60-67 Inches	7	15.9

Table 2: Awareness on various aspects

Data	(Frequency) %	
Down syndrome is genetically transmitted to offspring	Yes: (44) 100% No: (0)	Awareness on causality
Down syndrome is related to mental retardation	Yes: (8) 18.2% No: (36) 81.8%	
This syndrome is life long	Yes: (39) 88.6% No: (5) 11.4%	Awareness on management
This syndrome is treatable	Yes: (13) 29.5% No: (31) 70.5%	
This syndrome is manageable	Yes: (44) 100% No: (0)	
I had some knowledge regarding Down syndrome before the birth of this child	Yes: (44) 100% No: (0)	Awareness on pregnancy outcome
I was informed by the doctors regarding Down syndrome, before the birth of this child	Yes: (2) 4.5% No: (42) 95.5%	
Pre-natal screening is possible	Yes: (4) 9.1% No: (40) 90.0%	

Table 3: Awareness on Life Expectancy

(Frequency) %			
Data	Agree	Neutral	Disagree
Child born with Down syndrome are weak at birth	(25) 56.8%		(19) 43.2%
Normal developmental milestone	(41) 93.2%		(3) 6.8%
People with down syndrome can live up to average age		(24) 54.5%	(20) 45.5%
Life expectancy can be improved with quality of life		(12) 27.3%	(32) 72.7%

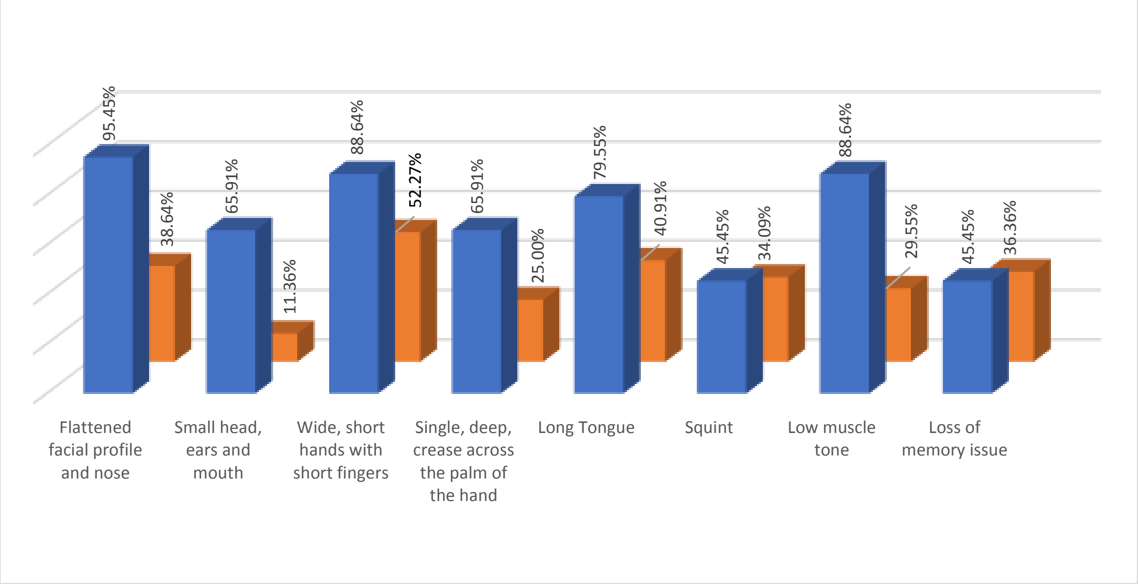

Figure 1: Knowledge regarding physical characteristics

DISCUSSION

Analysis of the questionnaire regarding the Knowledge of physical features of a child with DS showed that most 97% of the people were aware about facial profile and nose. 65.9% people responded positively regarding their knowledge about small head, ears and nose. 88.6% had knowledge about wide, short hands and short fingers. 65.9 knew about single deep crease across palm. 88.6 told that Down's children had long tongue. 45.5% knew they have squint. 88.6% have knowledge about loss of muscle tone. Regarding memory 45.5% knew they have poor memory while 93.2% knew they are slow learners. This is contrary to a study conducted by Levis DM in 2012 where participants were aware of facial features and few risk factors associated with DS[18]. However, in another study conducted in Saudi Arabia, important gaps in knowledge and awareness regarding DS were identified[20].

Results indicate that 100% knew about Down in our study. This is contrary to a study conducted by Alosaimi in Riyadh which showed that 64% of the mothers were unaware of the causes of DS[8]. Another study conducted by Lawal TA showed that mothers have poor level of awareness about birth defects[21]. Whereas Glimore et al reported awareness of DS as a chromosomal disorder among respondents[22].

In our study only 4.5% women were told before birth that their child has DS. Similar findings were observed in a conducted by Alosaimi where majority of respondents were unaware of the diagnosis of their child about DS before the birth. A variable response was seen in a study conducted by John ST that showed 46% of the mothers were aware of having DS child at birth despite concealment of fact by their physicians, where as 46% mothers were totally unaware of their child's condition delaying early intervention[12]. 9.1% women gave positive answer of possibility of pre-natal screening test. This is similar to a study conducted by Belachen et al where Women who have a child with DS have low awareness of existing screening tests for the condition[23].

When asked about whether children born with DS are weak at birth 56.8% mother agreed to it while 93.2% mothers agreed that they have normal developmental milestones. More than half of respondents had neutral response that People with DS can live up to average age. Similarly, 72.7% disagreed that quality of life can be improved along with life expectancy. A study conducted by Levis D et al reported misconceptions about risk factors, quality of parents life and DS life expectancy[18]. A study conducted by Huiracocha L reported that negative traits of society like bullying, humiliation and stigmatization towards families and children with DS hinder favorable attitude, multitude of efforts and hence development towards positive encouraging change in upbringing the differently abled child with DS[24].

CONCLUSION

Our selected candidates are mostly well aware of this condition but there is still the need for spreading further awareness to overcome the problems faced by the child and for their better management. Increased awareness along with early intervention not only improves the quality of life but is also crucial for the physical and mental well being of Down syndromes.

Conflict of Interest: All authors declare no conflict of interest.

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Ethical and Consent to participate: Informed verbal and written consent was taken from the institutes and the parents, we maintained the anonymity of the participants.

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