



# Insights into The Prenatal and Postnatal Knowledge & Critical Observations of Parents with Children who Have Down Syndrome: A Single Center Study

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## ABSTRACT

**Introduction:** Down syndrome (DS) is one of the many genetic syndromes commonly confronted by pediatricians. These people commonly share a few features which include a low level of muscle tone (hypotonia), learning disabilities, and abnormalities of the cranium and facial parts.

**Aims and Objectives:** To assess the pre and postnatal knowledge, of parents with Down syndrome children.

**Place and Duration of study:** This descriptive cross-sectional study was conducted in Shifa International Hospital (SIH), by Final year medical students, in association with the Cytogenetics Department. between December 2019 to September 2021

**Material and Methods:** All the children diagnosed with Down Syndrome at the SIH and their parents were invited to participate in our study. Convenience sampling was used depending solely on the number of parents who had accepted our invitation. A total sample size of 35 was included, of which 15 agreed to participate. A detailed questionnaire having both close ended and open questions in English and Urdu was filled out by those parents who consented. Data was entered and analysed using SPSS version 24.0.  $p$ value  $\leq 0.05$  was taken as significant.

**Results:** Our results showed that 11 of the 15 DS children i.e., 73.3% were male and 4 out of 15 i.e., 26.6% were females. None of these children had siblings with the same disorder. 13 of the 15 cases (86.6%) were diagnosed post-natally only 2 cases (13.3%) pre-natally. All the cases i.e., 100% were diagnosed by a pediatrician and none were diagnosed by an obstetrician. Surprisingly increasing age of either parent seemed to increase the incidence of DS offspring. 60.0% (9 of the 15) of parents did not know the condition could be diagnosed prenatally. While 10 of 15 (66.6%) of them were aware of its general complications. Answering open ended questions parents identified the lack of awareness, special educational institutes, and presence of communication gap between them and doctors as some of the factors that hindered the welfare of DS children.

**Conclusion:** Only one parent knew about DS complications pre-natally and only a few were aware that a pre-natal diagnosis could be formed. Postnatal knowledge of parents was sketchy also, especially regarding the future complications faced by DS children, Parents critically observed insufficient public awareness, societal taboos, paucity of local DS data and prenatal medical DS training, and consequently lack of governmental support required by special needs DS children in our country.

**Key Words:** Down Syndrome, Pediatrics, Lack of awareness, Pakistan, Learning Disabilities

## INTRODUCTION

Down syndrome is one of the many genetic syndromes commonly confronted by pediatricians<sup>1</sup>. The Down syndrome population commonly shares a few features which include a

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low level of muscle tone (hypotonia) which manifests at an early age, learning disabilities, and abnormalities of the cranium and facial parts. Individuals may experience a plethora of physical deformities like micrognathia, eyes becoming slanted, abnormally low muscle tone a single palmar crease, nasal bridge flattening out, and macroglossia<sup>2</sup>.

Genetic counseling is rarely done in many countries; then, it becomes the job of a pediatrician to break the news to the families and explain the diagnosis to them<sup>3</sup>. A few factors add to the difficulty in explaining the diagnosis to the family of the diagnosed child, such as giving birth to an unhealthy child, remaining in constant tension to fulfill the necessities of the child, and being anxious about the future holds for the child<sup>4</sup>. Studies suggest

that parents of Down syndrome children who had an eminent understanding with the healthcare professionals exhibited a more optimistic approach towards the care of the infant; parents were fonder of their babies<sup>5-7</sup>. Due to successful social support in the medical treatment, it has prolonged life expectancy for a Down syndrome child; the average life span has improved to 55 years in developed countries<sup>8</sup>.

We aim to assess the knowledge, attitude, and practice of parents having children suffering from Down syndrome- e.g., how well are they aware of this medical condition, how they cater to their needs, or the problems/ challenges faced while handling them, and other arising situations.

### MATERIAL AND METHODS

This descriptive cross-sectional study has been conducted in Shifa International Hospital, by Final year medical students, in association with the Cytogenetics Department. An IRB approval was obtained from Shifa Tamir – e- Millat numbered IRB#206-696-2019 dated 10 August 2019. Convenience sampling was done. The absence of any prior data enlisting total Down syndrome cases in the country was a hindrance. We identified all the kids diagnosed with Down Syndrome between December 2019 to September 2021 and their parents were invited to participate in our study. Of the 35 parents offered only 15 consented to do so. We used a detailed questionnaire in English and Urdu to be filled out by participants who consented to participate. All the assessable factors that needed validation or rechecking were taken from a previous study conducted in Turkey.<sup>9</sup> We translated the questionnaire into Urdu to make it comprehensible for native people who do not understand English to their full potential. The questionnaires and consent forms were made available via both hard and soft copies to participants who could opt for whichever was feasible. It was also aimed at including a maximum number of recruits in the study. Parents were informed about the nature of the study and informed consent was taken. The questionnaire had a combination of close-ended and open-ended questions, which helped us explore opinions about the options we offered as well as discover new ideas and concerns. The questions covered socio-demographic information (Table-1), details of time and method of diagnosis, and knowledge of the complications of the condition (Fig-2,3) as well as reasons for the lack of facilities for such kids and suggestions from parents for improving these kids' say in society as discussed later. All patient

information was kept confidential under double password-protected files, which were only accessible to the focal persons conducting the study. The participants had the liberty to leave the study whenever they wanted.

Statistical analysis was done using SPSS version 24.0. pvalue  $\leq 0.05$  was taken as significant.

Socio-demographic characteristics	N=15	(%)
<b>Gender of the child</b>		
Female	4	26.6
Male	11	73.3
<b>Mother's age at the childbirth (year)</b>		
<25	0	0
25-35	2	3.31
>35	8	53.3
<b>Mother's educational level</b>		
Illiterate	0	0
<8 years	0	0
$\geq 8$ years	15	100
<b>Father's age at the childbirth (year)</b>		
<25	0	0
25-35	4	26.6
>35	10	66.6
<b>Father's education level</b>		
Illiterate	0	0
<8 years	1	6.6
$\geq 8$ years	14	93.3

Table-1: Socio-demographic characteristics of the children and parents

### RESULTS

Our results were analyzed using Google Forms response analysis and SPSS 22, which showed that people are still reluctant to talk about the subject as only 15 of the 35 consented to participate in the study. 11 of the 15 (73.3%) were males and 4 of the 15 (26.7%) were females. Further illustrated in Fig-1 by DS male to female preponderance. 15 of the 15 cases (100%) were diagnosed post-natally and by a pediatrician and none were diagnosed by an obstetrician.

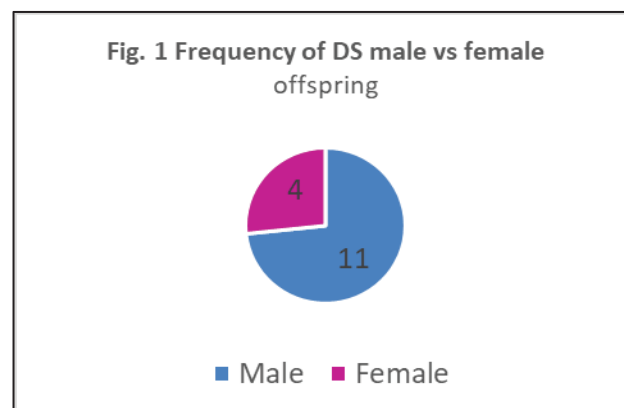
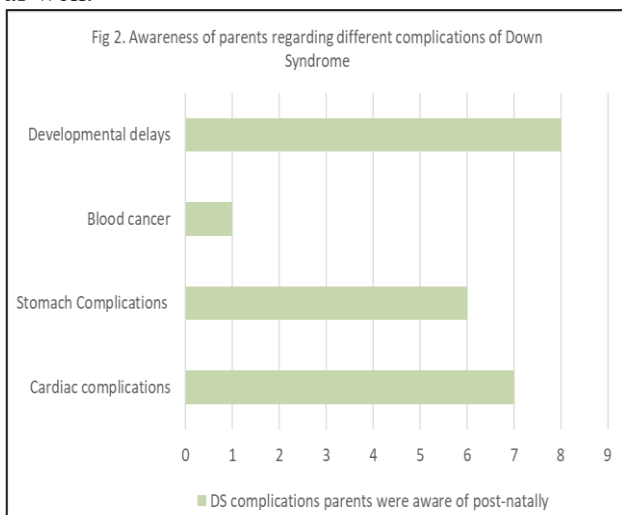


Fig-1: Frequency of DS male & female

There was a diverse level of education of parents, the mothers education ranging from Grade V up to M.Phil. level and that of the fathers ranged from matriculation up to M.Phil. level and also included a doctor. Of all the participants 1 of the 15 (6.67%) had a history of prior miscarriage and 14 of the 15 (93.3%) did not have a history significant for miscarriages. When the parents were probed about the awareness, understanding complications of Down Syndrome, despite local literature showing 88.2% of Down Syndrome with Congenital Cardiac Defects presented in infancy<sup>15</sup>, only (46.67%) parents 7 of the 15 knew about the risk of cardiac complications, 6 of the 15 (40%) knew about stomach issues, and most alarming was the fact that only 1 of the 15 (6.67%) knew about the risk of blood cancer in people with Down Syndrome. This is according to another study in Pakistan is that the proportion of DS-acute leukemia was found to be 1.84% among all cases of pediatric acute leukemia<sup>16</sup> details of which are which is depicted in Fig-2.

Lack of awareness among parents, insufficient special educational institutes to cater to the needs of kids with Down Syndrome, and communication gap between parents and doctors were identified as some of the major factors that hinder the welfare of DS children in Pakistan. When asked about avenues that could help such children contribute to society and lead a meaningful life, ideas including awareness campaigns, special education centers, government funds for these kids' welfare needs, the inclusion of kids with special needs along with other kids, so the society can better understand them and a multifunctional society with all types of kids can be formed, a helpline or correspondence unit to guide parents about the welfare, needs, and complications of the condition, were brought under the limelight as well.



**Fig-2: Awareness of Parents regarding different complications of Down Syndrome**

## DISCUSSION

Down Syndrome is the most common chromosomal abnormality prevalent in the world. A study in Norway, reported in the local population, the incidence of trisomy 21 was 0.19%.<sup>13</sup> Despite this, it is a social stigma in Pakistan. Parents are reluctant to voice their opinions about it, majorly due to its association with "bad luck" and that society would not accept these babies as "normal beings". This hesitancy can easily be noticed when parents were asked to consent and share their thoughts on this topic. One such research was conducted in Pakistan to gather views of parents with Down Syndrome kids, regarding the condition. Only 30 consented to share opinions while 9 did not<sup>10</sup>. Similarly in our study, only 15 out of 35 total parents were comfortable talking about this topic, while the remaining 20 parents refused to discuss the situation. This reluctance highlights the fact that such medical topics usually have a small sample size and that is why are still not thoroughly explored. Considering the lack of research on the subject a registry to study the the true prevalence, associated morbidities, and health-related quality of life (HRQOL) of these individuals and their families are not well documented, especially in low- and middle-income countries such as Pakistan is being proposed<sup>14</sup>.

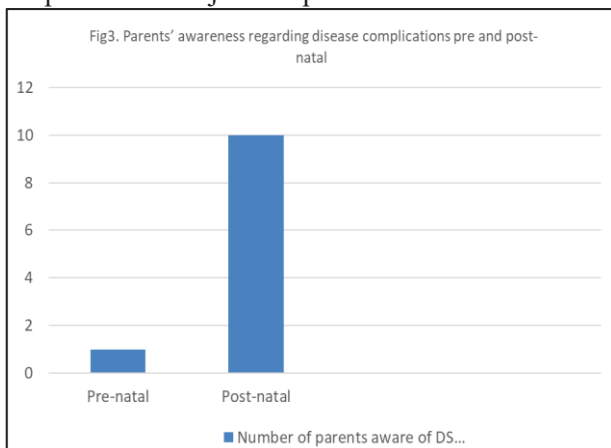
Advanced maternal age is one of the major predisposing factors to Down Syndrome. Thereby, if a mother is conceiving at a later age, the risk will multiply at age 25, it is 1 in 1250; at age 40, 1 in 100<sup>11</sup>. In our research, it was not only the advancing maternal age (age range from 28 to 37 years old) that was a risk factor but so was the increasing paternal age (ranging from age 29 to 49 years old). No one has commented on the fact that how paternal age alone could be a risk factor as well. Similar concerns and a probable explanation has been presented in a study the US, pointing out that the paternal age has also an effect on trisomy 21, specifically during male aging, when there is higher risk of chromosomal breaking in spermatozoa.<sup>17</sup> Our study highlights this important aspect which could be unveiled in future research.

While diagnosing the condition of Down Syndrome, it is the role of obstetricians to screen for Down Syndrome prenatally with the help of serum markers and ultrasound nuchal translucency scan, amniocentesis or chorionic villus sampling, according to the gestation<sup>12</sup>. An anomaly scan is usually done at 18-22 weeks of gestation to rule out cardiac, gastrointestinal, or spinal defects<sup>12</sup>. In our research, only 33% of parents knew about this scan

or were offered aptly. Early detection of anomalies can help in genetic counseling of the parents timely. Despite the major role of obstetricians, to our surprise, it was pediatricians who diagnosed all the cases in our research, thus owing to the alarming lack of knowledge among parents regarding the condition especially pre-natally, refer to Fig-3.

A similar trend has been noticed in other research as well<sup>3,10</sup>. However, one research conducted in Turkey reflected that still 37% of babies were diagnosed with the help of obstetricians<sup>9</sup>.

Genetic counseling is a vital part of diagnosing and preventing Down Syndrome. Parents must be guided about every aspect of the disease, so be it the negatives: fetal complications, birth defects, developmental delays, as well as the positives: availability of medical and surgical treatment and therapies. In our research, 67% were informed about genetic counseling. 80% were even instructed about karyotyping. 67% were aware about the general complications associated with Down Syndrome. It was a welcoming sight to see genetic counseling being offered to parents in Islamabad. It helped them understand the disease and plan future management accordingly. Such practices should be applied throughout Pakistan including public sector hospitals and not just the private centers.



**Fig-3: Parents awareness regarding pre and post-natal DS disease complications**

It is not uncommon to witness a plethora of complications associated with a Down Syndrome baby. These complications range from physical, mental, emotional, and biological aspects. Knowledge regarding complications of Down Syndrome was asked from the parents to see how educated they about the diagnosed topic were. Unfortunately only 47% of parents knew that there would be developmental delays; 20% were aware that a baby can be physically disabled, 20% knew it was not an infectious disease while 33% did not know that this was a genetic disease. A study

conducted in Turkey showed that 42% of parents were informed about the developmental delays, physical ailments, and nature of the disease<sup>9</sup>. Less than half (40%) of parents in our research realized that further medical disorders could later show up in life, for example, blood cancer, low IQ, low immunity, cardiovascular anomalies, and early dementia<sup>12</sup>. This necessitates how crucial it is to educate parents and make them fully understand the condition of their baby right from the beginning.

We questioned the parents what the factors could be causing hindrances in the progression of Down Syndrome children, and how they wanted to improve the condition of Down Syndrome babies in Pakistan. The results of the study revealed that parents believed the lack of knowledge regarding the topic to be the single most important factor resulting in stigma around this medical topic. They want to be educated more about the condition through optimistic doctors who refrain usage of negative terminology. Doctors would also guide about specific health issues and caring techniques according to the growing age and development of the baby in simple words, that is Urdu and any native language they are fluent in. This would also aid in bridging the communication gap causing any sort of obstruction in the knowledge of parents. They reckoned that no proper funding has been done by the government to meet special needs to cater to these babies. Proper teaching schools with elucidated syllabi should be set for them in the near future. To the popular belief that "these children cannot perform daily life duties or would underperform any given task", parents demanded equality in life and job opportunities like any other normal child would attain.

Our study took place in a well-established private hospital where a majority of parents had the means to afford and are well-off. So similar study can be conducted in government hospitals; this way true representation of the perception of Down Syndrome in the Pakistani population could be studied, and not just the financially stable and educated parents. The practice of obstetricians and pediatricians in government hospitals can also be researched, and then we could compare it with private hospitals.

Our research was limited by the small sample size due to the complexity and stigma revolving around the topic. It was conducted in a single private hospital confined to the province of Islamabad Capital Territory (ICT). The limited use of open-ended questions in gathering data restricted parents from voicing their opinions to their full potential. The limitations of our study were mainly a small sample size, which is insufficient to depict an entire

population, and added to it, the stigma associated with the condition makes it harder to convince people to enroll in the study. Future research should overcome these limitations to get to the depth of the subject and dig out information that is a better depiction and more beneficial to society.

### CONCLUSION

Parents expressed a significant lack of pre and post natal DS awareness. Only 1 parent knew about DS complications pre-natally and a few were aware that a pre-natal diagnosis could be formed. Postnatal knowledge of parents was sketchy also, especially regarding the future complications faced by DS children. An increase in the incidence of the DS children with increasing maternal and paternal age, was an important finding. A significant lack of prenatal medical training related to this subject was highlighted by the parents as only paediatricians diagnosed these cases postnatally. Parents drew concern regarding ignorance of society towards DS, alarming stigma related to the subject and government apathy towards their needs. All the parents felt a lack of facilities for these kids' needs was a key factor causing hindrance for them to excel and overcome obstacles to contribute positively to society.

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