Pakistani mothers' and fathers' experiences and understandings of the diagnosis of Down syndrome for their child

Kiran Jan Ahmed · Mushtaq Ahmed · Hussain S Jafri ·
Yasmin Raashid · Shenaz Ahmed

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Abstract Down syndrome (DS) is a relatively common chromosomal condition, which can be diagnosed prenatally. However, little is known about the diagnosis of the condition in developing countries. This qualitative study explored parents' experiences of the diagnosis of DS in Pakistan. Fifteen mothers and fifteen fathers of children with DS had semi-structured interviews, which were analysed using thematic analysis. All the parents received their child's diagnosis after birth, ranging from the postnatal period to 7 years of age. Parents recalled receiving little or no information at the time of diagnosis, leading to misunderstandings about the cause and nature of their child's condition. Some parents referred to their child being "Mongol" and were unaware of "Down syndrome" as the more appropriate term for the condition. Use of such terms for DS restricted parents' ability to source further information about the condition. Many parents showed poor understanding of the aetiology and prognosis of the condition. Improved training for healthcare professionals in recognising key features of DS in the neonatal period or in early childhood could enable earlier diagnosis of the condition. In addition, provision of accurate information in a

sensitive manner following diagnosis could enable parents to optimise their child's well-being.

Keywords Down syndrome · Diagnosis · Mothers · Fathers · Pakistan

Introduction

Down syndrome (DS) is a developmental condition commonly caused by the chromosomal anomaly Trisomy 21. The physical and non-physical features of DS are variable and unpredictable; one of the most significant impairments is a degree of learning disability. Pakistan is a country with high levels of learning disabilities associated with congenital conditions. DS is a main contributor to these levels of impaired mental development, where 1 in 300 live born have the condition (Gustavson 2005), although these figures are based on a study conducted in one city in Pakistan with a small birth cohort of 1,476 children.

Unlike many Western countries, Pakistan does not have a national screening programme for DS; prenatal testing for DS is available but only through private healthcare (Rahman and Obaid-ur-Rahman 2005). Due to the high costs associated with private healthcare, most women are unable to receive this testing; furthermore, around 65 % of the population live in rural areas where availability and access to prenatal testing for DS is limited (Government Of Pakistan 2009; Rahman and Obaid-ur-Rahman 2005). Therefore, the majority of children with DS in Pakistan are likely to be diagnosed postnatally.

Healthcare in Pakistan is provided by both public and private sectors, 25 and 75 %, respectively (Nishtar 2006). Public healthcare provision is inadequate and usually utilised by people from low socioeconomic backgrounds (Nishtar 2006). Private healthcare provision varies in standards of care from highly reputed and expensive hospitals in urban areas to

K. J. Ahmed
School of Medicine, University of Leeds, Leeds, UK

M. Ahmed
Yorkshire Regional Genetics Service, Leeds NHS Teaching
Hospitals Trust, Leeds, UK

H. S. Jafri · Y. Raashid
Genetech Laboratory, 146,1 Shadman Jail Road, Lahore, Pakistan

S. Ahmed
Leeds Institute of Health Sciences, University of Leeds, Leeds, UK

S. Ahmed (✉)
Leeds Institute of Health Sciences, School of Medicine, University of
Leeds, 101 Clarendon Road, Leeds LS2 9LJ, UK
e-mail: s.ahmed@leeds.ac.uk

small clinics run by unskilled personnel in both urban and rural areas (Malik 2011). The health system in Pakistan is poorly regulated and lacks specialist services for people with disabilities. We are not aware of any national or local guidelines in Pakistan for healthcare providers on managing the diagnosis of Down syndrome, of any written information about the condition for parents or of any *regular* care for children following the diagnosis. Medical students and paediatricians are trained to diagnose Down syndrome, but diagnosis by non-specialist doctors is likely to depend on them recognising the features of the condition, and by paediatricians is dependent on them coming into contact with the child. Therefore, a child can go years without receiving a diagnosis. In Pakistan, the institutional lack of recognition of Down syndrome as a condition requiring early diagnosis and the lack of a healthcare pathway are key challenges in improving the diagnosis of Down syndrome.

There is a dearth of literature on parents' experience of their child's diagnosis of DS in Pakistan. An awareness of this diagnostic process is essential for informing any necessary changes to improve clinical practice, training for healthcare professionals (HCPs) and information for parents in Pakistan and potentially in other developing countries, and in preparation for any future national screening programme for the condition. The aim of this study was to explore parent's experiences of the diagnostic process for DS for their child and their understandings of the information received upon this diagnosis.

Method

In-depth interviews were conducted by authors 2, 3 and 5. All the authors are of Pakistani origin and are multilingual in English, Urdu and Punjabi. Authors 1, 2 and 5 are based in the UK, and author 3 and 4 are based in Pakistan.

Participants

Thirty participants were recruited from a centre in Pakistan, which provided services for children and young adults with intellectual disabilities (referred to from here on as the Centre). The Centre was located in a large city in Pakistan where private prenatal screening for DS had been available for over a decade, and all the parents in this study lived in urban areas. The participants were all parents of children with DS—15 mothers and 15 fathers, representing 19 families (both parents of 11 children and either the mother or father of eight children). Both parents were included to ensure that any gender differences in parental experiences were captured. Purposive sampling of the parents of children with DS registered at the centre was conducted for gender, education, social background, parental age and the gender and age of child with

DS. Thirty-nine potential participants were approached and 30 agreed to participate. All the participants were Muslims and ranged in educational attainment, household income and employment status. Participants' age ranged from 37 to 61 years, and the ages of their children ranged from 2 to 30 years.

Procedure

The study received ethical approval from the University of Leeds Ethics Committee and all participants gave written consent to participate. The interview guide was developed to explore parents' experiences of having a child with DS, particularly the diagnosis of the condition. The interviews were conducted in 2008, at the Centre by multilingual researchers (in Urdu or Punjabi), separately with all the participants. The interviews lasted between 30 and 80 minutes, were audio recorded, translated into English and transcribed. The two transcribers were earlier career researchers based in Pakistan who were fluent in Urdu, Punjabi and English. Authors 1 and 5 assured the quality of the translation and transcriptions by listening to audio-recorded interviews and reading the transcripts simultaneously.

Thematic analysis was used to identify and interpret patterns of meaning within the qualitative data (Braun and Clarke 2006). A deductive approach was initially employed to organise and analyse data according to themes identified a priori, followed by an inductive approach to identify emerging themes. Data analysis also involved consistent cross-referencing between the participants for comparisons based on gender, age and educational status. All the data were analysed by the first author, who discussed the themes with the last author, then refined and discussed them again to ensure consistency in interpretation of the data. Differences by educational attainment and gender were explored, but not found.

Results

Experiences of delayed diagnosis

All the parents confirmed that their child was diagnosed with DS after birth. The time of the diagnosis ranged from the postnatal period to 7 years of age. A minority of participants said their child was diagnosed within a week after birth, where DS was recognised immediately, while the majority of children were diagnosed months or years later. Table 1 shows parent's level of education, the age at which the child was diagnosed with DS, the age of the child at the time the parents were interviewed, and the length of time between the diagnosis and interview, which ranged from 1 year and

Table 1 Parent's level of education; age of the child at diagnosis and at interview, and length of time between diagnosis and interview

Parent	Education(mother/father)	Age of child at diagnosis	Age of child at interview (years)	Length of time between diagnosis and interview (years/months)
F01 & M01	Master's degree/bachelor's degree	11 months	15	14/1
F02 & M02	GCSE level equivalent/no educational qualifications	At birth	4	4/0
F03 & M03	GCSE level equivalent/a level equivalent	4 months	2	1/8
F04 & M04	GCSE level equivalent/a level equivalent	3 months	7	6/9
F05 & M05	GCSE level equivalent/no educational qualifications	7 months	10	9/5
F06 & M06	GCSE level equivalent/bachelor's degree	At birth	11	11/0
F07 & M07	Master's degree/no educational qualifications	2 months	7	6/10
F08 & M08	Bachelor's degree/no educational qualifications	1 year	10	9/0
F09 & M09	Bachelor's degree/bachelor's degree	1 year	30	29/0
F10 & M10	Master's degree/no educational qualifications	4 months	20	19/8
F11 & M11	GCSE level equivalent/no educational qualifications	Few days old	8	8/0
F12	Bachelor's degree	At birth	8	8/0
F13	No educational qualifications	8 months	11	10/4
F14	Bachelor's degree	7 years	16	9/0
F15	Bachelor's degree	1 year	8	7/0
M12	Bachelor's degree	2 months	20	21/10
M13	No educational qualifications	2 years	16	14/0
M14	Master's degree	1 week	23	23/0
M15	A level equivalent	At birth	22	22/0

F&M indicates parents of the same child

F father, M mother

8 months to 29 years. During the analysis, no differences were found between parents' experiences of the diagnosis of DS and the information they received by parent's educational level or the lengths of time between their child's diagnosis and the interviews.

Many of the parents reported that the diagnosis occurred by chance: whilst visiting a doctor for seemingly unrelated health issues, such as, pneumonia, jaundice, viral infection, chest infection; whilst visiting the doctor for themselves and accompanied by the child; or when the child was receiving routine healthcare:

Father 4: A doctor came to the school giving some injections on some campaign, so I took her there. He looked at her and immediately said that she is Mongol.

Other diagnoses were made when parents took their child to a doctor specifically because of their concerns about their child's physical and/or mental development. The main concern for which parents consulted a doctor was absent or delayed speech development, usually around the time that a child would be expected to speak:

Mother 5: When he was 18 months, he did not speak, so I took him to the doctor. My other children started to

speak at around 1 year old, but he didn't so I was worried.

Father 8: ...he did not do things like normal children. His teeth developed late, he started sitting late, he started crawling late, I mean he did all things late.

Some parents also mentioned facial features as another reason for consulting a doctor:

Mother 6: Her tongue was very thick...her tongue was under her teeth...She looked different, but I said that it doesn't matter as it happens sometimes. She was weak... down's babies have round eyes, thick lips and body.... We took her to our family doctor and he wrote in her notes that the baby is a Down's baby.

Overall, diagnosis of DS was usually made by chance or due to parent initiated investigations based on their instincts and beliefs about the child's health and that their child was not "normal".

Knowledge of the name of the condition

Surprisingly, many parents were unaware of or could not remember being told the name of their child's condition.

Father 4: No, they didn't tell me the name of the disease... they just told me that it is from birth and cannot be cured and that it is genetic.

Mother 7: They didn't tell us the name... they just said he is mentally weak.

It may be that some of the doctors were aware of the condition but unaware of the name, or just unaware of DS. This issue is exacerbated by the fact there is no term for DS in the Urdu language. Furthermore, a term often used by parents to refer to the condition was "Mongol" and a number of parents remembered being told this term at the time of diagnosis:

Father 10: The doctor told us that he is retarded and he is Mongol.

Mother 4: No, they told us it is a Mongol child and they didn't tell us anything else.

The use of incorrect terms for the condition, i.e. Mongol, is likely to have restricted parent's ability to source further information about the nature of the condition, its treatment and the availability of health and social support. For example, a father explained how he researched his child's condition on the Internet using the term Mongol, but found information about the Mongol race instead and linked this to the condition that his child had:

Father 6: The doctor told me that your daughter is Mongol...I did research for this on the Internet and came to know that this is an ancient race, which looks like the Chinese. There was this race called Mongols, which lived near China. Their noses are flat and they have small eyes.

Understandings of Down syndrome following diagnosis

The three key messages that parents understood upon diagnosis of DS were that the condition is lifelong, incurable and results in delayed mental development. When questioned about how they felt at the time of receiving the diagnosis, parents talked about the challenges of each of these aspects. For example, the shock of finding out DS is a lifelong condition led to parents concerns about their child's independence in the future:

Father 7: The doctor told me there was a problem with the genes and my child would always be like this... I was depressed because the child would be like this forever... All we see is darkness. I mean what will he do? He remains dependant on others forever.

Parents' fears about whether their child would ever be able to achieve independence were based on their perceptions of

their child's poor education, job and marriage prospects as indicated by the doctors. Educational success was highly valued by parents as a means for leading an independent life, but following the diagnosis of DS, parents were worried about the impact of delayed mental development on their child's educational achievement:

Mother 4: (Interviewer- what did they tell you about the child at diagnosis?) Nothing. They only said she is mentally weak... I am worried as she cannot speak well, she cannot learn or get an education. It is necessary to be educated for survival in society.

Parents were also concerned about how delayed mental development would make it challenging for their child to get a job and financially support themselves. They hoped that their child would find some means of earning a living and lead a normal life by getting married:

Father 8: The doctors said that he would be slow mentally... I hope that he will learn some technical work to help him live his life independently.

Father 7: They said he is not normal and his development is slow... It is obvious that he will not become independent, so how will he get married?

Parents' ultimate concern regarding their child's lack of independence was who would care for their child after the parents died:

Father 1: No one knows about life and death. We have no answers to what will happen to him after we die.

Mother 4: I pray that I live long enough and can care for her.

When asked about what they were told at the time of diagnosis, none of the parents mentioned anything about the potentially shorter lifespan of their child with Down syndrome. Instead, their concerns about who would care for their child after their death suggested that they were unaware of the impact of Down syndrome on their child's lifespan.

Although parents understood DS was lifelong and incurable, they showed a limited understanding of the symptoms of the condition. Many parents said that they were not given much information about the clinical nature of DS. The main health implication that the majority of parents took away following the diagnosis was that their child would have delayed mental development. Surprisingly, parents rarely mentioned any physical features of DS. Children with Down syndrome are usually visibly different from parents and siblings, and physical features are the earliest indicators of the condition, whereas developmental milestones usually become apparent later on in the child's life. Only one parent mentioned concerns about a protruding tongue, one mentioned upward

slanting eyes and one mentioned delayed developmental milestones such as crawling and sitting, whereas, all parents put most emphasis on delayed mental development, particularly speech and language difficulties. This may be because parents were not informed about physical features related to DS upon diagnosis, or their children had mild physical features of DS but pronounced delayed mental development, or parents placed more emphasis on the mental development features of DS:

Father 15: He is mentally retarded comparing to other children.

Mother 5: He is mentally weak... he does not speak

Furthermore, it may be that parents simply did not realise that these physical features were early indicators of their child having a condition that may require medical attention:

Mother 6: She looked different, but I said that it doesn't matter as it happens sometimes.

None of the parents mentioned receiving any written information about DS. Instead, all of the parents recalled verbal information provision at the time of diagnosis. This may have led to hindering their understanding of DS. Some parents mentioned features of DS, but they did not attribute these to the condition. Instead, they attributed other explanations for these features. For example, some parents believed their child's speech difficulties were due to having small vocal cords or falling as a child:

Father 1: He has trouble talking properly, so I think that he fell down when he was younger when he was playing in the street.

Father 8: When he was about 4 or 5 years old, I saw that his vocal cords are very small so I thought about looking for someone to fix this so he could start speaking. But I can't find this sort of treatment.

Understandings of the cause of Down syndrome

Many parents said that doctors did not explain why their child had DS. Parents' recall of their conversation with doctors gave the impression that doctors themselves lacked understanding of the aetiology of DS. For example, some parents said that doctors told them that DS may run in their family and that they were questioned on whether they had a family history of DS. Other doctors suggested there may be a link with consanguinity by questioning parents' relationship to each other:

Mother 4: They said I had a Mongol child but didn't tell me why she is Mongol. They just kept asking me if I had married into the family, a relative.

Father 2: They didn't tell me how it occurs; they said that it might be in the family so they asked me about my family background. They also asked me if I was married to my cousin.

Parents showed misunderstandings about the causes of DS that resulted in feelings of guilt and upset, which made it more difficult for parents to cope with their child's diagnosis. For example, many parents believed that the condition may have been caused by taking certain medications or being stressed and tense during the pregnancy:

Father 7: I think it might be because of her reaction to a drug (medication) she took during pregnancy.

Mother 4: I took some medicine or some injection or some tension, so it is because of me.

Overall, parents felt that further progression in research and knowledge was needed to help them to understand and care for their child with DS:

Father 14: ...we do not know what the extent of his disease is... so much growth of knowledge is possible in this field. It is lacking right now. We need this knowledge to help understand our child's limits and capabilities to help solve his problems.

Discussion

All the parents in this study received their child's diagnosis after birth and were given little or no information at the time of diagnosis. They also had misunderstandings about the cause and nature of their child's condition. Most diagnoses were made months or years into their child's life. The main trigger for seeking a diagnosis was parents' perceptions of delayed developmental milestones, particularly delayed or absent speech, hence late indicators of the condition. Early detection of DS would allow early intervention and treatment provision, which could optimise the child's prognosis. Physical, occupational and speech therapy could help children with DS achieve developmental milestones with minimal delay (Hines and Bennett 1996; Majnemer 1998; Skotko et al. 2009). However, our study shows that parents in Pakistan are highly unlikely to obtain treatment and support for a child with DS during a crucial, early developmental period.

Furthermore, late diagnosis could have a negative psychological impact on parents. Studies demonstrate that parents who are uncertain of the cause of their child's delayed mental development suffer more emotional strain, regret and worry, in comparison to parents who have a diagnosis of DS for their child (Lenhard et al. 2005). Also, diagnostic uncertainty can lead to long-term negative psycho-emotional burden for

parents (Lenhard et al. 2005). However, parents with a definitive diagnosis can experience similar emotions to parents of non-affected children, possibly because DS is a common genetic condition with a fairly good prognosis (Choi et al. 2011; Devlin and Morrison, 2004; Lenhard et al. 2005). Early diagnosis can also reduce misdiagnoses and provision of incorrect information by HCPs (Poehlmann et al. 2005).

Early detection and diagnosis of DS in developing countries could be achieved by implementing national prenatal screening programmes. These could be similar to those in Western countries (Ekelund et al. 2008; National Screening Committee 2007), enabling parents to plan and prepare for their child both emotionally and practically (Poehlmann et al. 2005; Skotko et al. 2009). For example, parents could begin to identify support services and address and adapt their lifestyles. Prenatal diagnosis would also allow parents to make informed reproductive choices. Although there are religious issues relating to the provision of prenatal screening and termination in a country like Pakistan, such services are acceptable and currently available privately (Ahmed et al. 2013; Jafri et al. 2012). In the absence of a national prenatal screening programme for DS and to prevent delayed diagnosis, HCPs in Pakistan could be trained to recognise and diagnose phenotypic features in the neonatal period (Devlin and Morrison, 2004; Majnemer 1998; Skotko and Bedia, 2005). Furthermore, HCPs should be encouraged to consider a diagnosis of mosaic DS for children with unexplained delayed mental development, because phenotypic features are less commonly displayed in cases of mosaic DS (Devlin and Morrison, 2004).

Our findings show that the majority of the parents were unaware of the name of their child's condition and many referred to their child as 'Mongol'. Research in Western populations shows that parents find the term 'Mongol' insulting, arousing feelings of disgust and anger (Skotko and Bedia, 2005), and HCPs should avoid using it, adapting a more compassionate and respectful approach to discussing the diagnosis (Berg et al. 1969; Cunningham and Sloper 1977; Skotko and Bedia, 2005; Skotko et al. 2009). Overall, in Western clinical practice, the term is perceived as derogatory and offensive, and its use has been abandoned for many decades. Also, the use of this term alone by Pakistani parents is likely to restrict their ability to source information about DS. For example, searching the internet for the term 'Mongol' provides little information about DS, whereas searching for 'Down Syndrome' provides a plethora of useful information, including the nature of the condition, treatment and care options, and support services. It is unclear why HCPs in Pakistan use the term 'Mongol' for 'Downs syndrome'. Further research on HCPs use of this term could help develop materials to train HCPs about the benefits of using the correct term.

Our findings suggest that parents lacked understanding of key features associated with DS and they attributed other

explanations for their child's symptoms. Knowledge and understanding of the clinical features of DS is key in determining whether and how the parent seeks health and social care for their child. Upon their child's diagnosis, parents should be given information about symptoms and health complications of DS so that they can recognise and understand these features and seek appropriate support and treatment where necessary for the optimal well-being of the child (Skotko et al. 2009). Such information provision could be improved by ensuring HCPs in Pakistan are trained with up to date, accurate knowledge of DS and are able to communicate this in a sensitive manner, where parents can understand the implications for their child (Choi et al. 2011; Ferguson et al. 2006; Skotko et al. 2009). There are challenges in providing information about DS because of the variability of the symptoms and severity of the condition. Nevertheless, parents should be given a basic overview of DS with an explanation of its variability and directed towards sources of further information to suit their needs (Skotko and Bedia, 2005; Skotko, 2005; Skotko et al. 2009). The development and provision of written information for parents is also essential because they are unlikely to take in all the verbal information provided upon diagnosis due to their emotional state (Cunningham and Sloper 1977; Skotko and Bedia, 2005).

Parents in our study had limited information and understanding, and misconceptions about the causes of DS. They often formulated their own explanations, leading to feelings of self-blame and guilt about their child's condition. Misconceptions were reinforced because HCPs asked them misleading questions about their relationship to their spouse. Such questioning suggests that HCPs had a limited understanding of the aetiology of DS themselves. Training for HCPs should enable them to accurately communicate the risk factors for DS and the possibility of future children having the condition. Research shows that parents' desire for information on the causes and transmission of DS and the risks to their current and future children (Lenhard et al. 2005). Providing parents with this information may help them to understand and accept their child's condition, and may prevent unnecessary fear and concerns regarding any future pregnancies (Lenhard et al. 2005; Skotko et al. 2009).

As a retrospective study, this study is subject to recall bias, because parents were asked to recall their version of events leading to their child's diagnosis of DS years after the actual diagnosis. The reliability of the findings is dependent on the parents' memory of the information provided to them at the time of their child's diagnosis. Given that some parents may not be able to recall all details of what occurred at the time, this is a limitation of the study. Although it is possible that parents may have forgotten key information about the diagnostic process or information provided, many parents in this study appeared to recall the events clearly and with ease. This accurate, vivid and complete recall of the diagnosis can be

attributed to flashbulb memory (Conway et al. 1994; Finkenauer et al. 1998; Skotko and Bedia, 2005; Skotko et al. 2009). Interviewing parents within the first year of their child's diagnosis would have been preferred in order to avoid any potential recall bias; however, such a study would be impractical in Pakistan due to the lack of a general pathway for diagnosing children with DS, as demonstrated by our findings. Also, our study focuses on parents' experiences of the diagnosis of DS. These findings have relevance to other conditions where early diagnosis could have similar advantages for families, such as, Klinefelter syndrome (Bourke et al. 2013). Therefore, further research is needed on parents' experiences in Pakistan for the diagnosis of other conditions.

In conclusion, the parents in our study demonstrated that the diagnosis of DS in Pakistan may be delayed into childhood, where parents need to actively pursue a diagnosis based on their concerns about their child's development. Furthermore, upon diagnosis, parents were provided with insufficient or misleading information. Improved training for HCPs in recognising key features of DS could enable earlier diagnosis of the condition. In addition, provision of accurate information in a sensitive manner following diagnosis could enable parents to optimise their child's well-being. Overall, there is a need for national guidance on the diagnosis of DS and how to manage individuals with the condition.

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Conflict of interest Kiran Jan Ahmed, Mushtaq Ahmed, Hussain Jafri, Yasmin Raashid and Shenaz Ahmed declare that they have no conflict of interest.

Compliance with ethics guidelines All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (University of Leeds Ethics Committee) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all patients for being included in the study.

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