

Immediate and Delayed Psychosocial Effects of Down's Syndrome on Parents from Diagnosis to the First Post-Natal Year

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ABSTRACT

Down's syndrome is a common condition seen in society, with 1 in 1000 babies in the UK being born every year. Although seems common and known by a large proportion of the population, the effects on the family remain fairly hidden. Six major studies have provided evidence for how parents feel at the immediate stage of diagnosis and how their feelings evolve as their child develops. The theories of social acceptance and guilt have been applied to down's syndrome to help explain how parents come to term with their child's diagnosis and how the psychology of disability effects parents on a daily basis. Main findings include; parents having feelings of shock and disbelief at the time of diagnosis, parents would have liked to be informed differently to how they were and parents often receive most of their effects from third-parties through the form of stigma or prejudice. These effects should be highlighted to ensure they are more well-known in society so parents can be supported and are made aware of how to overcome them from other's experiences.

Keywords: Down's syndrome; Social relation model; Parental opinion; Stigma; Immediate effect; Delayed effect

INTRODUCTION

Representing the most common human autosomal aneuploidy, down's syndrome is a congenital condition associated to chromosome 21. Cytogenetic forms of down's syndrome trisomy's include; free, mosaic and Robertsonian translocation. Of these, Robertsonian translocation (2%-4% of cases) is the only form with an inheritance link. The remaining are nondisjunction (free) and cell division malfunction (mosaic). In all cases there is a resultant overproduction of chromosome 21 gene products causing the phenotypic abnormalities seen [1].

Down's syndrome effects 1 in 1000 babies (UK) equating to approximately 750 babies born a year. The average down's syndrome patient has four carers, of which three are family members, this highlights that the majority of the impacts is carried by family members. It's important understanding the familial effects of down's syndrome in order to best support them in the future [2].

MATERIALS AND METHODS

My main search was on Google Scholar where I used Boolean gates searching for "Down's syndrome AND effect on families". Filtering this to no earlier than 2018 revealed 15,500 articles, of which I used six main sources based on their relevant applicability and attainability. I've also utilised papers older than this, justified by them detailing core principles or first-hand accounts. I feel chronological bias isn't significant in the circumstance of down's syndrome as there has been little if no development in its treatment and every patient shows differing severities regardless of when they were born. I also searched for studies through PubMed for observational studies. Although randomised control trials would be preferred, it's ethically and physically impossible, so whilst interpreting this evidence I've taken into account bias and commented on this where appropriate [3].

I aim to develop a further understanding of how family members react and develop with a down's syndrome child and if this has any effect on them. I have concentrated on the effect on parents as including siblings wouldn't only bring about differing

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views which could complicate conclusions but uncover a breath of evidence too large to comment on in this paper [4].

RESULTS AND DISCUSSION

Immediate effect

There is little literature detailing parental opinion on having a child with down’s syndrome, with data concentrating on the patients themselves. Making this information known can help families have a predetermined understanding of what to expect and what is “normal”. Human nature allows us to assume what parents will feel when receiving the news of a down’s syndrome diagnosis, for example; shocked, upset and annoyed to name a few, however there is a lack of evident research showing this [5].

This has a correlation with the social relations model. This model depicts a statistical approach to studying how people perceive others, encompassing the effects of the perceiver, target and relationship. Simply establishing the child as the target and the parent as the perceiver, the relationship between them can be studied. Referring to the theories of social acceptance and rejection, there is a continuum of which parents can lie on at the time of birth. This can range from toleration to actively pursuing the child and be furthermore applied to disability in a more in-depth manner. Parents can choose to except the diagnosis and carry on with life as normal, some may struggle with their child and rely on support and others may be proactive in finding more appropriate ways to cope on their own (Table 1) [6].

Table 1: Developmental milestones for children with down syndrome-pediastaff.

Milestone	Range for children with down syndrome	Typical range
Gross motor		
Sits alone	6-30 months	5-9 months
Crawls	8-22 months	6-12 months
Stands	1-3.25 years	8-17 months
Walks alone	1-4 years	9-18 months
Language		
First word	1-4 years	1-3 years
Two word phrases	2-7.5 years	15-32 months
Personal/Social		
Responsive smile	1.5-5 months	1-3 months
Finger feeds	10-24 months	7-14 months
Drinks from cup unassisted	12-32 months	9-17 months
Uses spoon	13-39 months	12-20 months
Bowel control	2-7 years	16-42 months
Dresses self unassisted	3.5-8.5 years	3.25-5 years

One paper written in 2011 has combatted the issue of little research with an observational study of parent’s perspectives. Although not the most current, this paper brings about credibility in how it was made. The surveys, questionnaires and initial pilot study were approved by the University of Boston Review Board and the sample size of 300 families involving 4,924 individuals represented approximately 2% of the down’s syndrome population, a sizable coverage for a national study. However, participants were selected from the same area. Consequently, it’s feasible to believe that they had similar environments (healthcare pathways and socioeconomic

background) and this could explain the strength of the data that arose. Overall, I am confident in its findings, detailed in the following [7].

It is surprising to discover that many parents suffer from an amnesic effect on learning their child’s diagnosis. It’s hard to distinguish why this happens, but shock, disbelief and confusion are named emotions related to this. Specifically 18% of parents can’t remember what was first said to them regarding their diagnosis. Although not a particularly high figure it’s significant in the fact that most people would be expected to remember when they were told potentially life changing news.

It can be noted however, that the diagnosis hasn't changed the parent's views of their child. 99% of respondents expressed they loved their baby, 97% were proud of them and only 4% regretted having them. It can be drawn upon that parents are more effected by the news of a diagnosis rather than having to live with it. For example, once the diagnosis is accepted, parents have found their own ways to endure any complications and as the child gets older, they become more experienced in this [8].

If the news of a diagnosis provides the highest level of psychosocial effect on the parent, it's interesting to uncover why this is. The discussion surrounding how parents are told about their child's diagnosis has considerable bias but could be an independent variable and should be considered by all health professionals giving this information [9].

Delayed effect

The proceeding months and years after diagnosis are when psychosocial impacts can be appreciated to a greater degree. Firstly, loneliness can be a factor to consider. The effect on family has been described by Janet Carr in her book "down's syndrome: Children growing up". It uses data from 29 mothers, this is a good sample size considering the research follow up of 20 years, who were asked the same questionnaire at determined intervals about bringing up a child with down's syndrome. Its main aim was looking at distortion of opinion over time, however it also provides reliable information for opinion at the time of 6 months post birth. The study proved that in the first 6 months from birth mothers can become lonely, with 21% of mothers agreeing they had felt this way. Rather impressively it found that all mothers to down's syndrome children had less individual "social time" (going out without the child) than the control group. This was a significant finding, with a P value of <0.01, meaning there is confidence this is true. This reinforces findings from Lonsdale's long standing study in 1978 where it was proposed that living with disabled children can cause isolation and loneliness and further illustrates its application to down's syndrome [10].

However, it must be noted that 74% of mothers disagreed that it was the child making them lonely. This could suggest other factors such as third-party stigma being the causal influence in these effects. Stigma can be defined as incorporating "labelling, stereotyping, separation, status loss and discrimination". Applying this to down's syndrome, many family's members report that they have been victim to "courtesy stigma". Courtesy stigmas as described by Goffman is "sharing some of the discredit of the stigmatized person to whom they are related". Many researchers have proven this true, including Jain's systematic review of case reports, where it was proposed the stigma of disability caused others to stay away from the family as a whole [12].

Interestingly Carr's research also concluded that those who claimed loneliness also scored higher on the Malaise inventory score (Malaise scores were used to "describe appropriate descriptions of stress"). This suggests that individual mothers tended to be affected differently as a whole in multiple aspects of life, rather than similarly in some respects and differently in others.

This could be correlated to how each mother copes; for example, someone overwhelmed is more likely to score worse in more aspects of life. The most important factor from a mother's point of view (excluding all relating to their child) was their health. Unfortunately, Carr's work uncovered that 24% of the down's syndrome mothers reported worsening health (based on a check up with a GP) compared to that of only 14% in the controls within the first 6 months. It should be noticed though, that with the right guidance and information, mothers of down's syndrome children can prosper and live with no personal adverse effects, as shown by the remaining 76%. It purely places emphasis on the fact that mothers should be aware of their own wellbeing as well as their child's [13].

Excluded from the studies above is how guilt had affected parents. Guilt has been agreed upon by theorists as caused by a social transgression and can present as self-disappointment. From this, the theory of guilt portrays the emotions felt when we feel like we've done something wrong. The guilt seen in parents and disability is the 3rd type, seen when you think you did something-whether this is true or not. It can produce the same feelings as if you actually carried out a guilt resultant act.

Qualitative data on this subject was somewhat easy to find, with many mothers questioning what they did in their pregnancy that could've influenced development. Quantitative data proved difficult but one paper in 2010 which interviewed 100 parents of down's syndrome and 100 controls used longitudinal studies to form comparative data. It found that 22% of mothers and 9% of fathers felt guilty about their child's disability. This paper is unique because it included the opinions of fathers who are routinely neglected in research. Again, the finding links to stigma and misunderstanding. Of all down's syndrome only ~2% can be inherited and the inheritance pattern of this is yet to be clarified. This highlights there is by no means need for parents to feel guilty as it's completely random and could happen to anyone.

CONCLUSION

Overall, it can be said with confidence that the diagnosis of down's syndrome will have some effect on the immediate family members. However, the severity of this effect is case dependent and relies on many confounding factors. There is a marked difference between the effects seen at different times of the child's life. Although different for every parent the immediate impact tends to focus on shock and the reality of the situation coming to life. After months and years have gone by this "new" reality isn't new anymore, the circumstance has been accepted and parents can live a normal life. The effects seen at these later stages involves coping with complications of down's syndrome and the challenges it brings in order to live as normal a life as possible.

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