Exploring the diagnostic odyssey of families affected by 22q11.2 deletion syndrome

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This study aimed to understand the experiences of parents and carers with regards to obtaining a diagnosis for their child. Five parents and carers of children with the disorder were recruited through support groups. Semi-structured interviews were conducted with participants to obtain qualitative data. These were then transcribed and analysed.

Three themes were identified:

1. Accessing support and navigating services
2. Psychosocial burden
3. Coping and adjustment

This study found that parents identified little benefit to traditional genetics services, other than as an information source. When accessing other services, participants were limited based on where they lived and their financial status.

Parents utilised social support networks, including family, friends and the 22q community. However, the impact of having a child with a rare disorder left them isolated as the divide between them and their peers widened. Women were disproportionately impacted, with both their social and professional lives.

Despite the psychosocial burden the diagnosis brought, parents were able to learn to cope and adjust to the new normal. This was achieved through both internal factors:

- An effective grieving process – often over many years
- Looking after themselves as well as their child
- Empowerment as a result of information gathering

And external factors:

- Good professional support
- A solid social support network

Some participants even finding positivity in the diagnosis and the person that it had made them into.

Overall, we found that parents need more support both in terms of care coordination and for their emotional wellbeing. Genetics professionals are effectively providing information, but not always addressing the needs of the parents in front of them. The mental health of the parent can be impacted by the diagnosis, and parents may need signposting to support for themselves, not just for their children.

Further research is also needed into disparities in support as, to our knowledge, there is no published data on this topic specifically for 22q11.2DS. Learning more about the patients who are missed by the social and medical care systems can provide invaluable information and is the first step towards being able to address these issues.