Exploring the journey to genetic services: A qualitative study of parental perspectives of children with rare disease

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Introduction

- Rare diseases are associated with extended diagnostic delays which are more financially and psychologically costly than that of more common disease.
- Most rare diseases (80%) are thought to be genetic in aetiology. However, uptake of genetic services is limited.
- Understanding the experience of the diagnostic journey for rare disease is essential to best facilitate access to and awareness of genetic services.
- The diagnostic experience remains unexplored in Queensland (QLD) and Victoria (VIC).

Aims

- Explore the experience of the diagnostic journey for parents of children with rare disease in QLD and VIC.
- Explore what it means to parents to receive a diagnosis for their child.

Methods

- Inclusion Criteria: Parents of children with rare disease with experience seeking a diagnosis for their child in QLD or VIC
- Semi structured interviews were used to explore the experience of the diagnostic journey in QLD and/or VIC
- Interviews were transcribed verbatim and thematically analysed.
- Recruitment text posted in the Syndromes Without A Name (SWAN) Australia newsletter and closed facebook groups

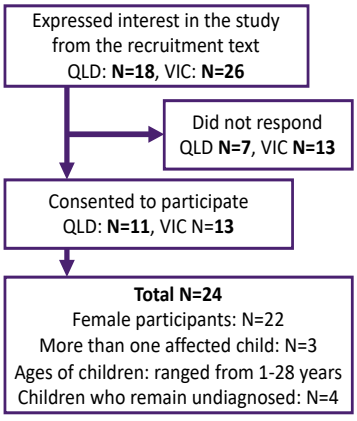

Figure 1: Recruited from VIC and QLD via Syndromes Without A Name Australia facebook groups and participant characteristics.

Results

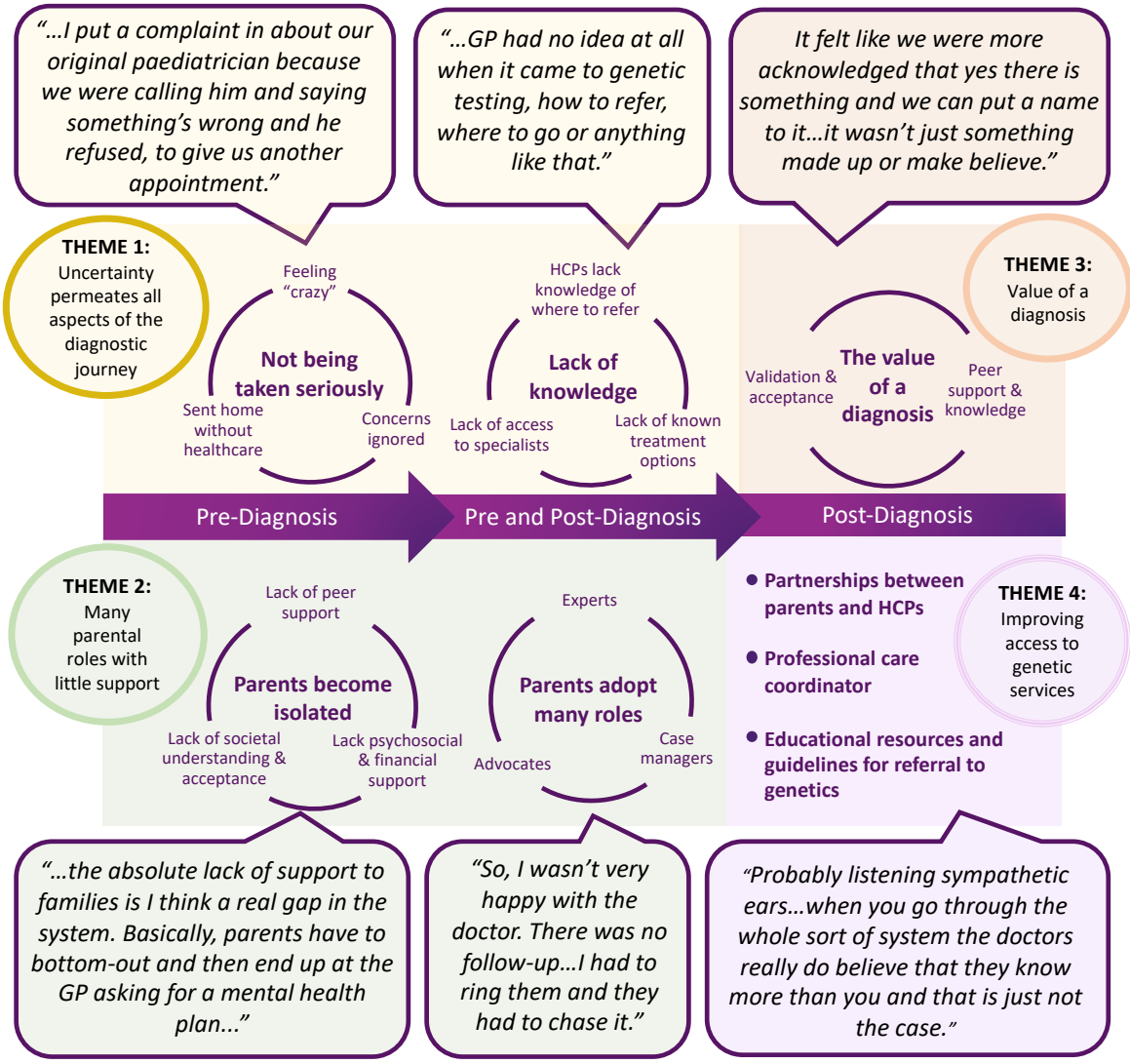

Figure 2: A visual representation of parental experiences with the diagnostic journey for rare disease pre-diagnosis, pre and post-diagnosis, and post-diagnosis. Themes and representative quotes are highlighted for each aspect of the journey. HCPs denotes "health care providers".

Conclusion

- Both parents and HCPs experience uncertainty through the diagnostic journey, and parents described having many responsibility for their child's care including the role of 'care coordinator', with minimal support.
- Parents reported that a diagnosis was validating and it allowed them to connect and engage in information sharing with parents who had similar experiences - "social precision medicine"
- We emphasise the call for:
  - Educational resources aimed at improving awareness of genetic services for non-genetic HCPs.
  - Support from a HCP to coordinate care, ensure consistency in and communication between healthcare teams and improve communication between parents and other HCPs.
  - Parent-HCP partnerships where HCPs acknowledge parental expertise and work with them to facilitate the best care for the child.

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