

Returning Individual Genetic Results to Research Participants: Experiences of Stigma in South African Families with Neurodevelopmental Disorders



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01 INTRODUCTION

Neurodevelopmental Disorders (NDDs) are a group of diverse disorders that manifest in childhood and result in deficits of gross or fine motor, cognitive, speech and social skills. Children with NDD's are often stigmatised as: **"Dangerous", "Mentally-ill", "Weird", "Violent", "Lazy", "Aggressive"**. Parents may be socially excluded, blamed and face status loss.

Due to the lack of genetic data in African populations, the NeuroDev Study was implemented. The NeuroDev Study aims to expand the phenotypic and genetic aetiologies of NDDs in South Africa and Kenya through large scale genomics research. Some research suggests that there is potential for genetic attribution to increase pre-existing stigma while others suggest that it may not increase stigma. This has remained understudied in African populations.

The role of Genetic Counsellors in addressing stigma in genomics research is also understudied and was thus of interest to the researcher.

02 OBJECTIVES

To determine:

- The experiences of parents or caregivers of NeuroDev research participants with stigma.
- The influence a positive genetic finding may have on stigma experienced.
- The role of Genetic Counsellors in addressing instances of stigma.

03 METHODS

Qualitative phenomenological study that used an Interpretive Phenomenological Approach (IPA). Purposive sampling along with semi-structured interviews were used to obtain results pertinent to the research question. The results were analysed using an IPA-tailored thematic analysis as seen below.

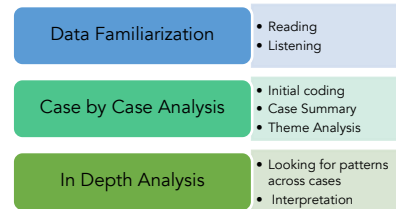


Figure 1: Methodological flow used in this study.

04 RESULTS

Parents shared experiences of **public, associative** and **internalised** stigma.

Table 1: Excerpts of stigma experienced by parents and caregivers of children with NDDs .

Stigma	Type	Description	Excerpt
Public	Mocking	People mock children with NDDs	"Mad" -P2/ "Sidalwa"-P1/ "Yeah you walking your dog"-P5
	Exclusion	Family members exclude son from social events	"Everybody's welcome except [son]"- P5
Associative	Blame	Nurse blamed mother for daughters behaviour. Her daughter has Autism	"Can't you control your child?"-P2
		Transport driver blamed mother for her daughters condition and hospital stay.	"You deserve this because you put your career before your family. Because you decided to have a child so late. This is clearly an act of God to punish you for"-P3
Internalised	Isolation	Participant felt alone since people don't understand Autism.	"There's nobody that understands what I'm going through, how I'm feeling"-P6
	Guilt	Participant felt at fault for her daughters condition	"I feel like it's because of me that she's this way"- P2

Case-by-case analysis illustrated that a **holistic framework** influenced the perception of a positive genetic result and subsequently, experiences of stigma.

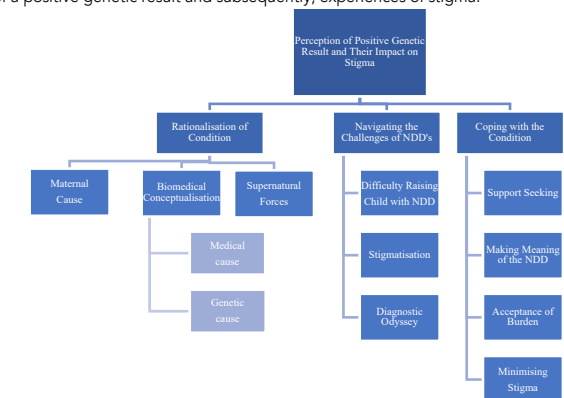


Figure 2: Framework that influences the perception of a genetic result.

A positive genetic result influenced internalised stigma however, it did not affect other forms of stigma. When the role of Genetic Counsellors was questioned, Genetic Counsellors were shown to positively impact genomics research and internalized stigma. Other forms of stigma were not affected

05 DISCUSSION/CONCLUSION

There is no **"one-size-fits-all"** in stigma experiences and how a genetic result influences these experiences. These concepts interact and intersect uniquely, resulting in individualistic experiences.

Coping mechanisms were shown to help parents face adversity and deal with stigmatising experiences. The genetic result only impacted internalised stigma experiences: removes responsibility.

Did not impact other forms of stigma: possibly due to lack of communication of positive genetic result, poor understanding of genetics or because they receive immense support from their families and community.

Genetic Counsellors play a role in reducing internalised stigma but no effect on public and associative stigma. Genetic Counsellors operate on an individual level rather than community level.

Genetic Counsellors were found to be a resource of information, provide a human element to genetic research and ensure continuity of care. Consequently, Genetic Counsellors are shown to play a crucial role in genetics research.

06 LIMITATIONS, FUTURE RESEARCH & IMPLICATIONS

Limitations

- Complexity of qualitative research for first-time researchers, selection bias, minimal comparative literature.

Future Research:

- Consider unique experiences of those not interviewed and the understanding of genetics and Genetic Counsellors among lay public.

Implications:

- Promote understanding of stigma among medical professionals, direct genomic studies in South Africa and promote inclusion of Genetic Counsellors in genomic research.

07 ACKNOWLEDGEMENTS

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REFERENCES

De Mini, V., Hoogenhout, M., Kikemoi, P., Kamuya, D., Eastman, E., Galvin, A., ... & Robinson, E., 2019. The NeuroDev study: Phenotypic and genetic characterization of neurodevelopmental disorders in Kenya and South Africa. *Neuron*, 101(1), pp.15-19.

De Vries, J., Landroo, G. and Workman, A., 2020. Stigma in African genomics research: Gendered blame, polygeny, ancestry and disease causal beliefs impact on the risk of harm. *Social Science & Medicine*, 258, p.113091.

Faure, M.C., Matshabane, O.P., Marshall, P., Appelbaum, P.S., Stein, D.J., Engel, M.E. and de Vries, J., 2019. Does genetics matter for disease-related stigma? The impact of genetic attribution on stigma associated with rheumatic heart disease in the Western Cape, South Africa. *Social Science & Medicine*, 243.

Matshabane, O.P., Campbell, M.M., Faure, M.C., Appelbaum, P.S., Marshall, P.A., Stein, D.J. and de Vries, J., 2021. The role of causal knowledge in stigma considerations in African genomics research: Views of South African Xhosa people. *Social Science & Medicine*, 277.

Parkevicz, I. and Smith, J.A., 2014. A practical guide to using interpretive phenomenological analysis in qualitative research psychology. *Psychological journal*, 20(1), pp.7-14.

Talbot, F., Bull, S., Farsides, B., Newport, M.J., Adeyemo, A., Rotimi, C.N. and Davey, G., 2009. Impact of social stigma on the process of obtaining informed consent for genetic research on podocriosis: a qualitative study. *BMC medical ethics*, 10(1), pp.1-10.

Serehuk, M.D., Corigan, P.W., Beed, S. and Olan, J.L., 2021. Vicarious stigma and self-stigma experienced by parents of children with mental health and/or neurodevelopmental disorders. *Community mental health journal*, pp.1537-1546.

Reddy, G., Fewster, D.C., Gurupath, T. Parents' voices: experience and coping as a parent of a child with autism spectrum disorder. *South African Journal of Occupational Therapy*, 2019 Apr;91(1):43-50.

Talbot, F., Bull, S., Farsides, B., Newport, M.J., Adeyemo, A., Rotimi, C.N. and Davey, G., 2009. Impact of social stigma on the process of obtaining informed consent for genetic research on podocriosis: a qualitative study. *BMC medical ethics*, 10(1), pp.1-10.