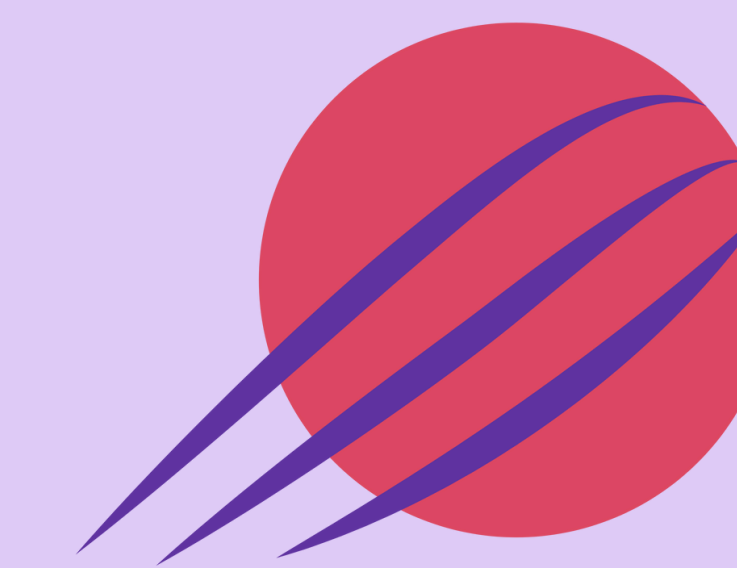




# From Deficit To Difference: Depathologizing The Language Of Genetics

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## Introduction

- Language can shape how individuals and their families perceive genetic variations, how healthcare professionals communicate diagnoses, and how society views genetic differences (Cho et al., 2023).
- Despite rapid advances in screening and healthcare, genetic differences—such as Sex Chromosome Aneuploidies (SCAs), the most common types of chromosomal variations (Tartaglia et al., 2020)— are still largely described in negative terms with words like “disorder,” “abnormality,” and “disease” shaping discussions.
- While depathologized language is increasingly recognized in fields like oncology and autism research, its application in genetics remains underexplored. A shift toward person-centered language may help improve well-being, reduce stigma, and promote more inclusive healthcare and educational practices.

## Methods

- Lived experience data is a valuable yet often underutilized resource in genetics research, offering insight into how genetic differences impact daily life and well-being (Kaur, 2025).
- This presentation shares preliminary findings from a qualitative analysis of publicly available podcasts (N=12) featuring individuals and parents of children with SCAs and Trisomy 21 (*The Chromodiversity™ Podcast*, 2022–2024).
- Using thematic analysis of interview transcripts, we explored how genetic language impacts participants' self-perception, experiences, and access to support across medical, educational, and social settings. Additionally, we reviewed existing literature on the impacts of clinical versus depathologized language.

## Results

### Psychological Impact and Identity

- *“Klinefelter syndrome didn't really sit with me... I'm more than a syndrome, and more complex than that.”* – **Person with XXY**
- *“I am not lazy. I am not a bad person. I have just this chromodiversity.”* – **Person with Mosaic Turner's**
- *“We're not a disorder... we're not a syndrome... we're people.”* – **Person with XXY**
- *“I don't like the word disease because it's not a disease at all. It's just a difference.”* – **Person with XXY**
- *“There's a lot of positivity that can be emphasized... with strengths like creativity, curiosity, perseverance, kindness, social intelligence, and fairness.”* – **Parent of child with XXY**

### Stigma and Dehumanization

- *“Your child is your child first. Just like when a child has cancer, you don't call them a cancerous child.”* – **Parent of a child with Down's**
- *“Historically, we've been kept in the shadows... a point of shame and a thing people don't talk about. We need to celebrate it now.”* – **Person with XXY**
- *“The more we're defined as a disorder or something that needs to be fixed... the more we're gonna have problems as a community.”* – **Person with XXY**

### Barriers in Healthcare and Education

- *“Why when a physical therapist or a school teacher or anyone meets my kid, they only know the diagnosis as per the textbook? Why are they not looking at my child first?”* – **Parent of child with Down's**
- *“I wanted the school to get to know my children for who they are and to see their strengths before planning for any challenges.”* – **Parent of children with XXY**
- *“[The diagnosis] was very clinical, very dry, no emotion, and no meaningful content that really helped understand.”* – **Parent of child with Down's**

## Discussion

- Findings highlight the negative psychological, social, and health impacts of overly pathological language. Participants expressed frustration at the sense of being reduced to a “syndrome” or medical label rather than seen as individuals, aligning with research showing a strong preference for more optimistic portrayals (Jaramillo et al., 2018).
- Findings also suggest that pathological framing of genetic differences reinforces stigma and may hinder access to appropriate healthcare and education. Research in other fields like autism and cancer genetics demonstrates that person-centered language can improve psychological outcomes and social integration (Bottini et al., 2024; Hunter et al., 2022).
- Multiple participants positively reframed the language surrounding their diagnosis and highlighted its importance. Strengths-based approaches are shown to improve self-efficacy and social-emotional skills in students (Thompson et al., 2021), suggesting that promoting individual strengths in children with genetic differences leads to better quality of life.
- While pathological language may be useful for purposes of clinical clarity or in securing funding, it should not dominate patient interactions. Trauma-informed communication and simplified language can improve understanding while minimizing harm (Schlub & De Deckker, 2024).
- Further investigation is needed to assess how person-centered language in genetic care influences long-term well-being.

Clinical	Depathologized	Example
Deficit Syndrome Disorder Patient Abnormality	Challenge Variation Difference Person Chromodiversity	<p><i>“Klinefelter is a natural variation in human DNA associated with unique strengths and differences.”</i></p> <p><b>versus</b></p> <p><i>“Klinefelter syndrome is an abnormal genetic disorder associated with multiple deficits and diseases.”</i></p>

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