



# Family Experiences and Attitudes About Receiving the Diagnosis of X & Y Chromosome Variations: Preliminary Results

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## Introduction & Background

- X & Y chromosome variations are the most common chromosomal abnormality, occurring in one out of every 300-400 births.
- Historically, many individuals remain undiagnosed during their lifetime, however increased utilization of cell-free DNA screening has increased the incidence of prenatal diagnosis of these conditions.
- In the pediatric setting, X & Y variations are diagnosed after genetic testing for unexplained symptoms, including developmental delays, behavioral disorders, learning disabilities, and abnormal pubertal development.
- Diagnoses are frequently incidental or secondary findings following genetic testing to confirm a suspected condition.
- Increased availability of clinical genetic testing means parents may receive their diagnosis from a provider other than a genetic specialist or counselor.
- The purpose of this project was to investigate how parents learned of the diagnosis, who supplied the information, how this information was received and their experiences with receiving support.

## Methods

- In 2018, a mixed-methods approach was used to elicit the viewpoints of individuals and parents of children with an X & Y chromosome variation on the experience of diagnosis and support needs.
- The study survey was modeled on a prior survey by Skotko et al. developed for the Down syndrome community.
- A web link to the anonymous survey was disseminated through AXYS listserv and associated social media platforms.
- The survey was conducted between October 2018 and February 2019 and open to all individuals and parents of children with an X & Y chromosome variation over the age of 16 and English-speaking.
- The survey was fielded through RedCap and remained open for 8 weeks in September and October 2018.
- Qualitative analysis of parents who have children with an X & Y variation explored the differences between parental reactions to a prenatal and postnatal diagnosis.

## Results

Overall, 323 Caregivers responded to the survey. Of the 323 respondents, 56 were not included in the analysis due to missing data. Characteristics of the remaining 252 respondents are shown in Table 1. Characteristics of the children with X & Y variations are shown in Table 2. Persons who disclosed the diagnosis are shown in Table 3.

Table 1. Parent Characteristics

Variable	Mean Age in Years	SD
Age at Child's Birth	33.4	5.7
	Number	Percent
<b>Ethnicity</b>		
African American	3	1.2
Hispanic or Latino	12	4.8
White	221	87
Asian	8	2.7
<b>Parent Education</b>		
Less than 8 <sup>th</sup> grade	2	.8
Finished 8 <sup>th</sup> grade	27	10.7
High School	138	54.8
College	69	27.4
Graduate School	16	6.3

Table 2. Child Characteristics

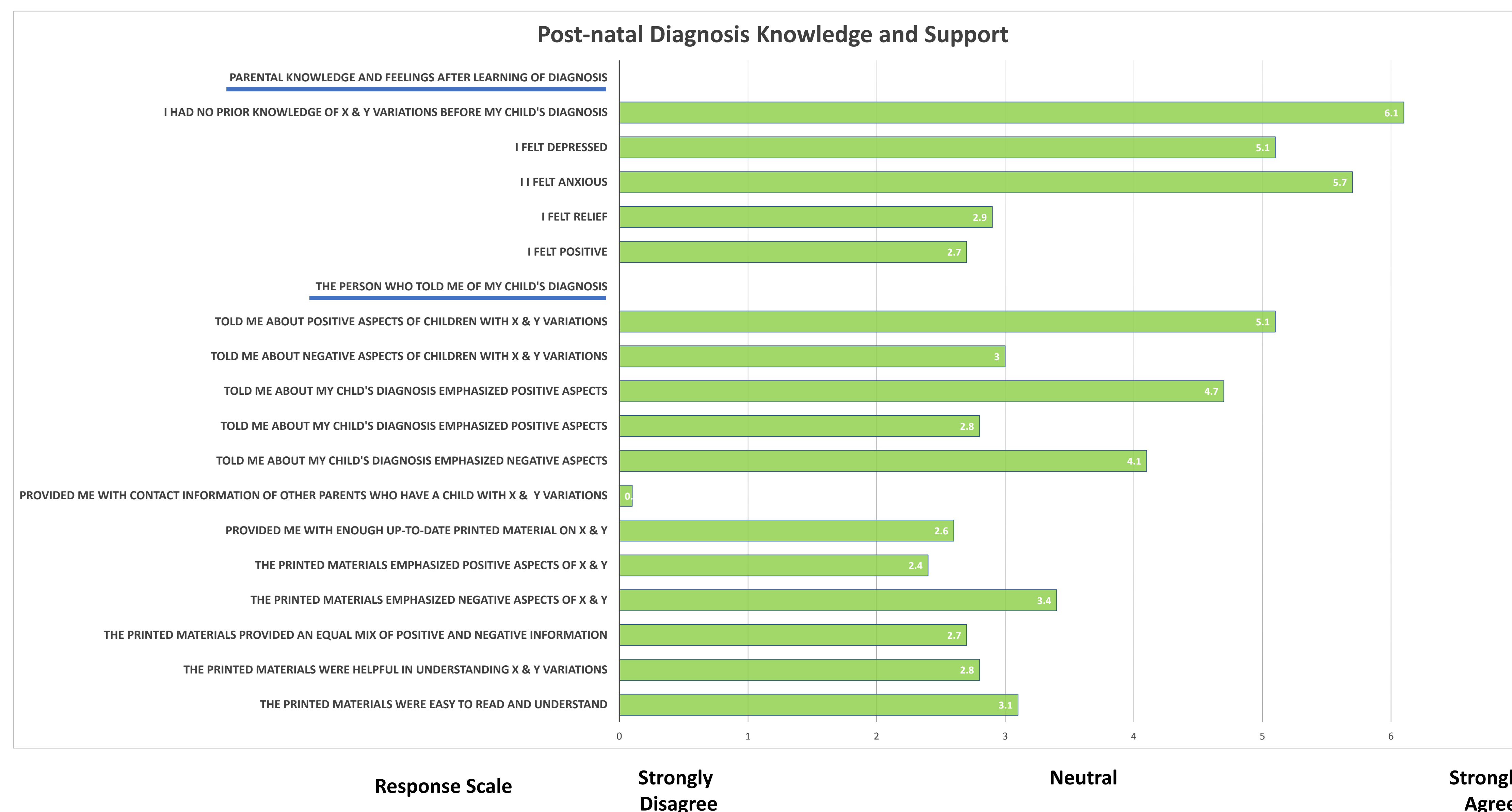
Variable	Frequency	Percent
<b>Biologic sex</b>		
Male	192	76
Female	60	24
<b>Gender Identity According to Parent</b>		
Cis-gender	251	99.9
Non-Binary	1	0.003
<b>Timing of Diagnosis</b>		
Prenatal	112	44.4
Postnatal	142	55.3
<b>Variation Type</b>		
47, XXY	117	46.8
47, XXX	51	20.1
47, XYY	37	14.6
48, XXXY	14	5.5
48, XYY	8	3.2
48, XXXX	4	1.6
49, XXXXY	9	3.5
Other	12	4.7

Table 3. Original Diagnosis Disclosure

Variable	Frequency	Percent
<b>Who Disclosed X &amp; Y Chromosome Diagnosis</b>		
Obstetrician Maternal Fetal Medicine Provider	54	17.6
Geneticist Genetic Counselor	82	26.7
Primary Care Provider	105	34.2
Specialty Provider*	41	13.4
Other**	25	8.1

\*\*Specialty providers include: Endocrinology, Cardiology, Urology, & Psychiatry

\*\*Other includes: Spouse, Child, Non-Invasive Prenatal Screening Laboratory



## Exemplary Quotes

- *"My pediatrician could have given me a list of 'what to do next' or insisted that I see a genetic counselor. It would have been nice to have someone help me put together a multi-disciplinary team to provide treatment for my son."*
- *"It was awful finding out the way we did in front of our son with only negative connotations."*
- *"It was over the phone and the doctor said she thought it was an error and she had never heard of it before. She thought lab made an error and apologized if our son had this condition."*

## Discussion

- Preliminary results demonstrate that parents who receive the diagnosis of an X & Y variation in a child receive this news from a variety of providers.
- Post-diagnosis support for parents is lacking in the delivery of accurate knowledge while providing necessary emotional support.
- There appears to be a general lack of adequate information available for providers and parents concerning what to expect during development and how to support a child with an X & Y chromosome variation.

## Conclusions & Recommendations

- Parents, patients and family members need to be included in the development of informational materials intended to help others understand issues and concerns about X & Y variations.
- Further research is recommended to develop interventions that prepare parents and families for receiving this diagnosis and for health care providers to disclose the diagnosis.
- Foundations in health care provider education as well as continuing professional education are needed to increase awareness of these conditions.

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