

Appendix

Table A: Coding Analysis

Code	Description	Sample Quote
Diagnosis	Providers' qualitative portrayal of condition; Individuals present at diagnosis; Reactions to diagnosis.	"From there they learned that's XXX, so they had me come in and speak with a genetics person there who just kind of—again I was very naive to the whole thing—just explained that if you have a chromosome disorder, it's the best one that you can wish for. That she'll be taller than average. She could possibly have speech and cognitive delays, and that's about it. I thought hey, no big deal. That's all the information that I had on it at that time. That's kind of where it started."
Pregnancy Termination	Parents' thoughts and attitudes toward; Provider discussion of; Discussion of with family/friends.	"When those results came in, the doctor called me on the phone, and he said that even though I'm at Week 24—'cuz [state] allows up to 24 weeks, where, I guess, you can abort, 'But you may wanna consider terminating the pregnancy. We can get around the [state] rule.'"
Emotional Responses	Includes positive and negative responses; Anxiety or stress	"Initially, it was a grieving process. We cried a little bit.

	<p>experienced following diagnosis; Concerns over finances, family dynamics, etc.; Seeking emotional reassurance, depression (meds) etc.</p>	<p>Initial questions were not necessarily, ‘What’s the prognosis?’ because, genetics, you can’t change. ‘What do we need to do? What do we need to put in place to make sure that he can be successful, not just in school, but, in life in general? What is going to—what are the typical characteristics of somebody with XXY?’ I think that was our biggest concern.”</p>
<p>Future Recommendations</p>	<p>Includes recommendations to society, medicine, educational system, other parents, desires for research, etc.</p>	<p>“I think a great way to start would be journal articles and not written for the average layman, written for the medical community, that you see—you have a—if you’re testing—for perinatal doctors for women over the age of 35, and they’re getting this news that maybe the best way to deliver it is not over the phone. Call them in. Call the husband in. Say, ‘This is what’s come up,’ and say this is not your area of expertise, but, ‘Here’s some studies.’”</p>

Interview Protocol for Parents

Thank you for agreeing to participate. Can you start by telling me a little about your family?

Prompts: How many children do you have? What are their names? How old are they?

Can you tell me a little about your son/daughter with [Klinefelter/XYY/Turner]?

Prompts: What do they like to do? What grade are they in? What hobbies do they have?

Part 1:

When did you find out about [child's name] diagnosis?

Prompts: Where were you? Who delivered the diagnosis? What did they say about [the condition]? Were you and your partner together? How did you feel? What thoughts were going through your head?

What did you do next?

Prompts: Where did you look for information? Who did you talk to? Did you feel supported?

What kind of conversations did you have with your family?

Part 2:

Did/How did the diagnosis impact your prenatal/neonatal care?

Prompts: Do you feel anything changed in your medical care? What medical advice were you given?

How would you say [child's name] diagnosis has impacted your relationship with doctors/nurses/the medical system?

Prompts: Where did you look for medical resources? What services were you offered? Do you generally feel that you've received enough or not enough medical support? Are there services you have not been able to find/access?

Thinking more generally, how well do you think individuals with [Klinefelter/XY/Turner] are supported by the medical system?

Prompts: What programs/services have been most helpful? Which have been least helpful?

Are there services you think should be available more broadly? What have you heard from others about their success in accessing services?

How accurate do you think people in the medical system's perception of [child's condition] is?

Prompts: How do clinicians' explain the condition? Do you think they generally put a positive or negative spin on the condition? What positive or negative interactions have you had with how people understand [child's condition]?

Part 3:

Do you think [child's name] diagnosis influences how s/he interacts with you and the rest of your family?

Do you think it influences how s/he interacts with her friends/schoolmates/peers? How would you say the diagnosis has impacted your family socially?

Prompts: What social support or services have you been offered/received? Do you feel that your family has received sufficient support? What support/services would you like to see implemented?

How would you say [child's name] feels about school?

Do you think the diagnosis has impacted how you and [child's name] interact with teachers/administrators/the educational system?

Prompts: Have special educational services been available? How effective/helpful would you say the services have been? What services would you like to see implemented?

How accurate do you think the general perception of [child's condition] is?

Prompts: What kind of things have you heard about [the condition]? Where do you think people get their information? How would you like to see the perception of [the condition] change?

Part 4:

What other areas would you like to see research in? Is there anything else you think we should talk about? Is there anyone else you think I should talk to?

Thank you for your time today. Please feel free to follow up with me if you have further questions.

Interview Protocol for Young Adults

Thank you for agreeing to participate. Can you start by telling me a little about your family?

Prompts: How many brothers/sisters do you have? What are their names? How old are they?

Can you tell me a little about yourself?

Prompts: What do you like to do? What grade are you in? What hobbies do you have?

Part 1:

When did you find out about your diagnosis?

Prompts: Where were you? Who delivered the diagnosis? What did they say about [the condition]? Were you and your parents together? How did you feel? What thoughts were going through your head?

What did you do next?

Prompts: Where did you look for information? Who did you talk to? Did you feel supported? What kind of conversations did you have with your family?

Part 2:

Did/How did the diagnosis impact your medical care?

Prompts: Do you feel anything changed in your medical care? What medical advice were you given?

How would you say your diagnosis has impacted your relationship with doctors/nurses/the medical system?

Prompts: Where did you look for medical resources? What services were you offered? Do you generally feel that you've received enough or not enough medical support? Are there services you have not been able to find/access?

Thinking more generally, how well do you think individuals with [Klinefelter/XY/Turner] are supported by the medical system?

Prompts: What programs/services have been most helpful? Which have been least helpful? Are there services you think should be available more broadly? What have you heard from others about their success in accessing services?

How accurate do you think people in the medical system's perception of [the condition] is?

Prompts: How do clinicians' explain the condition? Do you think they generally put a positive or negative spin on the condition? What positive or negative interactions have you had with how people understand [the condition]?

Part 3:

Do you think your diagnosis influences how you interacts with you and the rest of your family?

Do you think it influences how you interact with your friends/schoolmates/peers? How would you say the diagnosis has impacted your family socially?

Prompts: What social support or services have you been offered/received? Do you feel that your family has received sufficient support? What support/services would you like to see implemented?

How would you say you feel about school?

Do you think the diagnosis has impacted how you interact with teachers/administrators/the educational system?

Prompts: Have special educational services been available? How effective/helpful would you say the services have been? What services would you like to see implemented?

How accurate do you think the general perception of your condition is?

Prompts: What kind of things have you heard about [the condition]? Where do you think people get their information? How would you like to see the perception of [the condition] change?

Part 4:

What other areas would you like to see research in? Is there anything else you think we should talk about? Is there anyone else you think I should talk to?

Thank you for your time today. Please feel free to follow up with me if you have further questions.