

Summary from Kathleen Erskine:

Thank you to everyone who participated in this survey and for letting me hear your stories. Your insights were invaluable to the success of this project.

The purpose of this study was to create an educational booklet for parents to give to their daughter when they tell her about her Trisomy X diagnosis. In order to create this booklet, I created a survey for three separate groups of people: parents who had disclosed the Trisomy X diagnosis to their daughters, parents who had not disclosed the diagnosis, and women and teens with Trisomy X. After reading the scientific literature, I came up with the following list of possible discussion topics:

- Genetic causes or concepts
- Height
- Motor difficulties
- Learning disabilities and academics
- Speech and language delay
- Social challenges
- Behavioral or emotional difficulties
- Concerns about puberty
- Strengths

I also asked a few questions about general issues surrounding disclosure and advice to other parents or girls with Trisomy X.

Overall, 64 people completed the survey (26 parents who had disclosed, 32 parents who had not disclosed, and 6 women with Trisomy X). From the responses, the average age of disclosure was approximately 9-10 years of age, and was therefore made the target age group for the booklet. Below is a summary of the information included in the booklet from the responses to the survey:

- Genetic causes or concepts: explain chromosomes, how an extra X can happen, the extra X is not due to the mother's or girl's fault
- Height: she may be taller than other girls her age, the benefits of being tall
- Motor difficulties: difficulties with gross or fine motor skills, muscle weakness or tiring
- Learning disabilities and academics: Trisomy X is associated with learning disabilities, coping strategies, not using Trisomy X as an excuse for not trying in school
- Speech and language delay: importance of speech therapy, difficulties with self expression, finding the right words or remembering words, difficulties understanding others and understanding jokes
- Social challenges: behavior in large social situations or among friends, shyness, addressing the idea of feeling different while not wanting to be different
- Behavioral or emotional difficulties: specific behavioral or emotional challenges, managing anger and frustration, emotional coping strategies

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- Concerns about puberty- most people who responded said they did not address this issue as it pertains to Trisomy X, so it was not included in the booklet
- Strengths: specific strengths mentioned were sense of humor, creativity, love of animals, skills with younger children, athletic, artistic, and musical abilities
- Other topics that were included: not wanting to feel different, privacy of diagnosis, normalization of difficulties, variability of Trisomy X, not using Trisomy X as an excuse

Advice given to parents regarding disclosure includes having the conversation as early as possible, have simple explanations, and be honest but positive.

After finishing this project, I came to the following three conclusions:

- There is an overwhelming need for more research regarding Trisomy X.
- From all of the responses, a large number of parents weren't sure what they would discuss with their daughters.
- It may be helpful for girls with Trisomy X to have a way to communicate with each other or with older girls and women with Trisomy X.

Thank you again for participating. The booklet is available here:

<http://www.genetic.org/Portals/0/Articles/47,XXX/Guide%20to%20Trisomy%20X%20Booklet%20-%20Erskine.pdf>