

Why?

Most girls with Tetra X have three X chromosomes from their mother and a single X chromosome from their father. When a mother's eggs form, chromosome pairs usually separate so that each egg has 23 chromosomes, including a single X. When a mistake occurs in cell division, two X chromosomes can remain in the egg. If the same mistake is repeated at the next division, the egg can end up with three X chromosomes. Fertilised by a single X-carrying sperm, the egg will then develop into a Tetra X baby with four X chromosomes. Very occasionally, other processes may be involved, such as a failure of the X chromosome to separate in the normal way during cell division in the fertilised egg after conception.

Will it happen again?

Tetrasomy X is an extremely rare event and there are no reports in medical papers of families having either more than one daughter with Tetrasomy X or any other sex chromosome condition such as Triple X (having one additional X chromosome). However, women with Tetra X may run an extra risk of chromosome disorders if they have children. A discussion with a geneticist or a genetic counsellor will help to clarify the situation for you.

Prenatal diagnosis

Most pregnancies are normal and it is unusual for Tetrasomy X to be diagnosed before birth. However, occasionally there are signs that might suggest the need for a chromosome analysis. Some babies grow very slowly in the womb. Analysis of the chromosomes from either chorionic villus sampling or from amniotic fluid would normally show clearly the presence of the two extra X chromosomes.

Will a girl with Tetra X lead an independent life?

Many girls with Tetra X lead independent lives. Experience shows that some girls achieve full independence while others need some level of support and supervision.

Families say ...

... seeing her grow and surpass all expectations has made me realise her strength and potential - age 18.

... a caring, loving young lady, thoughtful in so many ways - age 19.

... an accomplished writer, she has written poetry and a five-chapter novel - age 20.

Inform Network Support



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<http://www.rarechromo.org/donate> Please help us to help you!

When you are ready for more information, *Unique* can help. We can answer individual queries and we also publish a full leaflet about the effects of Tetrasomy X. This information sheet is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. The information is believed to be the best available at the time of publication and the medical content has been verified by Dr Raoul Rooman, Paediatric Endocrinologist, University Hospital, Antwerp and by Unique's Chief Medical Adviser 2004

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Understanding Chromosome & Gene Disorders

Tetrasomy X



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What is Tetrasomy X?

Tetrasomy X (Tetra X) is a very rare chromosome condition in which girls have two extra X chromosomes, giving them four X chromosomes in all. Although girls with Tetra X are alike in certain ways, the extent of the effects of the extra X chromosomes can vary hugely. In some girls they are scarcely noticeable, while in others they cast a profound influence on the girl's life and her family.

These features are particularly common in girls with Tetra X. Not every girl will have all or even most of them but most girls will have some.

- **Slight delay in physical (motor) development**
- **Delay in starting to speak**
- **Some learning difficulty, usually no more than mild or moderate**
- **Above average height as an adult. This may follow slow growth as a baby and young child**
- **Possibly, increased vulnerability to emotional, social and behavioural stress**

How is Tetra X diagnosed?

A blood sample is taken. Cells are taken from this blood sample and treated so that the chromosomes can be examined under a high resolution microscope. The extra chromosomes can usually be clearly seen through a microscope.

How common is Tetra X?

This is not known, because there are almost certainly girls and women with Tetra X who do not know they have it. There is a growing worldwide community of over 60 women and girls who know they have Tetra X.

Development

Although girls with Tetra X do share similar features, the extent of the effects varies hugely.

■ Motor Development

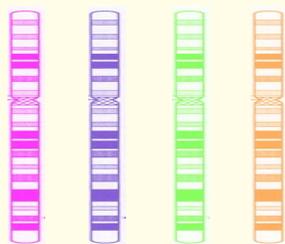
Girls are typically late in learning to sit, crawl and walk. Once they become mobile, most go on to play sports, games and to dance, but some tire easily and may lack stamina. For some girls, tight finger joints and a poor grip can make everyday manual tasks difficult.

■ Learning

Most girls have some level of learning difficulty although the range is very wide. Typically, difficulties are mild or moderate and girls will benefit from extra support with their learning. Some girls have a specific speech delay. Parental backing, setting realistic goals and educating girls in a supportive and encouraging environment can be transformative.

■ Speech

Speech delay is common and it is known that with too many X chromosomes abilities such as language and attention may be adversely affected. There is likely to be a particular problem with verbal skills, and in general verbal intelligence may be lower than non-verbal abilities (such as shape manipulation and understanding spatial relationships).



The typical karyotype is 48,XXXX

What is the outlook?

Your daughter's outlook is predicted best by the paediatricians who check her health in the early weeks and months of her life. Most girls and women with Tetra X are healthy and there are many adult women with the condition.

Medical conditions

- **Heart conditions** affect many girls but are often minor and may not need treatment. One girl in five has a more severe or complex heart problem and for them, surgery is usually completely successful.
- **Joints** Excessive or limited movement of large and small joints is common. Abnormal formation of the hip joints and club foot (talipes) can be found and some girls develop a curvature of the spine. Virtually all the orthopaedic problems typical of Tetra X can be successfully treated with splinting, casting or surgery.
- **Puberty** Half of Tetra X girls will have normal periods while the others will have menstrual irregularities and possibly underfunctioning or absent ovaries. Oestrogen treatment may be necessary to induce puberty and to prevent osteoporosis (brittle bones).
- **Fertility** A very few women with Tetra X are known to have had children. However women with irregular periods or ovarian problems will be less fertile. Girls with Tetra X may find it helpful to discuss their options with a geneticist or genetic counsellor if they are contemplating a pregnancy.
- **Behaviour** Some girls lack self confidence and are vulnerable to stress and anxiety. They benefit from a stable, loving environment that allows for any frustration caused by communication difficulties and for any teenage moodiness.
- **Psychiatric** Some girls do have bouts of aggressive and difficult behaviour and a few develop psychiatric symptoms. Because of the known vulnerability to stress, it is wise to seek counselling early. In *Unique's* experience behaviour management and medication are usually successful.

