Why?

It is most likely that your daughter has four X chromosomes from her mother and one from her father. When a mother's egg cells form, chromosome pairs usually separate to leave each cell with a single X chromosome. When a mistake occurs in cell division, two X chromosomes can be left in the egg cell. If the same mistake is repeated at the next cell division, the egg will have four X chromosomes. Fertilised by a single X-carrying sperm, the egg will then develop into a Penta X baby with five X chromosomes.

Every parent of a Penta X girl wonders: 'Why me?' In truth, this is a random event. Nothing you did before you were pregnant or during pregnancy caused Pentasomy X and there is nothing you could have done to prevent it.

Will it happen again?

Pentasomy X is extremely rare and chromosome tests on the parents usually show that both have normal chromosomes. In this case your chances of having another daughter with Penta X are no higher than for anyone else in the population. Very occasionally, a mother who has never suspected any problems before turns out to have a single extra X chromosome in some of her cells, and then her chances of having another affected daughter will be higher. As each family is unique, all parents of a girl with Penta X should have a chromosome study and a personal interview to discuss the findings with a geneticist or genetic counsellor.

Are Tetra X and Penta X the same?

No, girls with Tetra X have four X chromosomes and girls with Penta X have five. But there are many more girls with Tetra X than with Penta X and as they share similarities, families can learn from each other.

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.

Inform Network Support

Understanding chromosome disorders Unique

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G1 The Stables, Station Road West, Oxted, Surrey RH8 9EE, UK Tel/Fax: +44(0)1883 723356
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Tetrasomy and Pentasomy Support Group

www.pentasomy.com

Unique mentions other organisations' message boards and websites to help families looking for information. This does not imply that we endorse their content or have any responsibility for it.

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 Pentasomy 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

Rare Chromosome Disorder Support Group Registered in England and Wales
Charity Number 1110661
Company Number 5460413

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Pentasomy X

rarechromo.org

What is Pentasomy X?

Pentasomy X (Penta X) is a very rare chromosome condition in which girls have three extra X chromosomes, giving them five X chromosomes in all. Although girls with Penta X are alike in certain ways, the extent of the effects of the extra X chromosomes can vary hugely. In some girls the effects are not very noticeable, while in others they cast a profound influence on the girl's life and her family.

How is Penta 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 Penta X?

Medical researchers have suggested that Penta X may affect as many as one in 85,000 girls. In truth, no one really knows how common it is, because there are almost certainly girls and women with Penta X who do not know that they have this condition. *Unique* has 11 member families and a handful more families belong to the Online support group that you can find through www.pentasomy.com. Since 2000 this group has been active and very supportive.

Common features

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

- **Slow growth as a baby and young child**
- **Delay in physical development**
- **Delay in starting to speak**
- **Some level of learning difficulty**
- **Possibly, increased vulnerability to emotional, social and behavioural stress**

Development

No systematic survey of a large group of girls with Penta X has yet been carried out. This information therefore comes from *Unique*'s experience and from the descriptions of individual girls in the medical literature.

■ Growth

Your daughter will probably be small at birth and may be tiny, though in proportion. She may also put on weight very slowly in the first months. So long as she gains weight steadily and is well, she may not need any calorie supplements.

■ Mobility

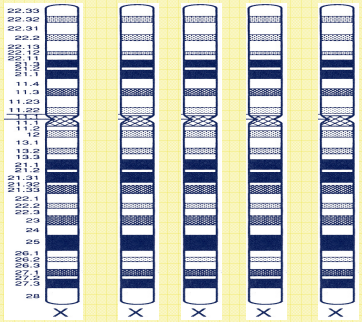
Many babies (though not all) are unusually floppy and quite a few girls have joints that are either unusually lax or stiff. This hinders their mobility and most are late to sit and walk – but the great majority get there in the end.

■ Learning

Girls with Penta X generally learn at a somewhat slower pace than other girls. Many are able to start their education in a mainstream school, but may later find their needs are better met in a special school.

■ Speech

Speech is usually delayed but girls communicate well at first with other means such as facial expression and vocal noises. Once first words emerge, articulation may be unclear and speech therapy is usually helpful.



The typical karyotype is 49,XXXXX.
Some girls have a mixture of cells with 48,XXXX and 49,XXXXX chromosomes.

Nine months old

The front cover shows a girl with Penta X at 3 years

Medical conditions

- **Heart conditions** are quite common, and your daughter will be very thoroughly examined. Most heart conditions are minor and resolve naturally with time, but occasionally girls have complex heart conditions that require surgery.
- **Joints** Stiff, clicky, lax or even dislocatable joints are a common feature and your daughter may need support for her ankles when she starts walking. Some girls also need their hip joints stabilised as babies. Exercise, swimming, physiotherapy and massage help extend the range of movement and ease any discomfort.
- **Kidneys and urinary tract** Your daughter will probably have a renal scan to check that her kidneys and urinary tract are healthy and functioning well.
- **Infections** It is possible that some girls with Penta X are especially vulnerable to infection and take longer than other children to recover from illness.
- **Teeth** Milk and permanent teeth usually emerge late and may have insufficient enamel. Girls should have good dental care, including fluoride painting if parents wish.
- **Constipation** has been noticed by some families. Simple steps such as encouraging drinking and high fibre foods may be a helpful precaution.
- **Puberty** This is not yet certain, but girls may well start puberty late and may experience some difficulties, such as scant or absent periods.