

Why did it happen?

In the great majority of cases, no cause is known and it should be regarded as an accident that happened in cell division in the process of making sperm or egg cells. Accidents such as these are not uncommon and they can affect children from all parts of the world and from all types of background. They also happen naturally in plants and animals. So there is no reason to suggest that your lifestyle or anything that you did caused it.

Can it happen again?

PKS has only been known to occur sporadically, so that the affected couple and other family members appear to be no more likely to have another child with PKS than anyone else in the population.

Diagnosing PKS during pregnancy

Pregnancy is usually uneventful but in some pregnancies with a baby with PKS, ultrasound scans raise concerns. In some cases, there is an unusually large amount of amniotic fluid (polyhydramnios). To make the diagnosis, the baby's chromosomes need to be analysed. An amniocentesis allows cells shed by the baby into the amniotic fluid to be examined. It is usually accurate, although diagnosing a mosaic condition is difficult because of the neighbouring populations of cells with a different chromosome make up. It is also not possible to distinguish mild cases from severe ones just by looking at the chromosomes.

What could happen next?

After diagnosis, regular checks will be kept on your baby's wellbeing. Sadly, some pregnancies with mosaic tetrasomy 12p end in stillbirth. Babies may also be born prematurely and need considerable support as newborns. Some babies do pull through but others do not.

Diagnosing PKS after birth

PKS is usually diagnosed after concern arises either over birth defects, appearance or development. Chromosome analysis from a blood sample will often show a normal result unless it is taken very soon after birth. Skin samples or from inside the mouth usually show the 12p chromosome for longer. This means that it can take months or occasionally years to reach the diagnosis.

Inform Network Support



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Websites, Facebook groups

<http://pkskids.net>

<http://pkskids.ning.com>

www.pkssitalia.org

www.facebook.com/pages/Pallister-Killian-Syndrome-PKS

www.facebook.com/pages/Sindrome-Pallister-Killian
(in Spanish)

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 answer individual queries and publish a full leaflet on Pallister-Killian syndrome.

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 content of the full leaflet on which this text is based was verified by Dr Michel Vekemans, Department of Genetics, Hopital Necker Enfants Malades, Paris, France 2004

and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, UK, Unique's chief medical adviser 2005

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Pallister-Killian syndrome

rarechromo.org



What is Pallister-Killian syndrome?

Pallister-Killian syndrome (PKS) is a rare chromosome disorder.

The genes that contain coded instructions to the body to develop and work properly are located on chromosomes. Normally there are 46 chromosomes in a cell, occurring as 23 pairs. Twenty-two of the pairs are numbered from 1 to 22, roughly from longest to shortest. The 23rd pair are the sex chromosomes. Each chromosome has a short arm ('p' for 'petit') and a long arm ('q').

People with PKS have two different lines of cells in their body. In one cell line they have a small 47th chromosome, called an isochromosome, composed of two copies of the short (p) arm of chromosome 12. In the other cell line they have the normal number of 46 chromosomes. This chromosome make up is called mosaic tetrasomy 12p.

Genes on the 47th chromosome cause PKS. However, the severity of the effects varies enormously. For reasons that are not yet well understood, some people have mosaic tetrasomy 12p without the distinctive features of PKS.

Main features of PKS

- Marked floppiness (hypotonia) in babies
- Developmental delay
- Learning difficulties
- Delay in developing speech or no speech
- In babies and young children, a distinctive appearance, with a high, rounded forehead, widely spaced eyes and a large mouth with a thin upper lip
- Sparse hair or bald patches around the temples, filling in by around 5 years, and thin or sparse eyebrows.
- Streaks or patches of darker or lighter skin, obvious at birth or emerging later

How did it arise?

In the process of making egg and sperm cells, chromosome pairs separate so that each cell receives 23 single chromosomes. A fairly common error is a failure to separate (non-disjunction), so the cell receives an extra chromosome 12. The 12p isochromosome is then created when one chromosome 12 splits horizontally at the centromere instead of lengthwise as usual. The long arm disappears and mosaicism arises soon after conception when the isochromosome is lost from one cell line.

Development

■ Growth

At birth, babies are typically normal or large. As adults, they are not unusually tall, though.

■ Learning

Most children will need support with their learning. The range of ability is wide and cannot be predicted before birth but will become evident as a child develops. From what we know today, it seems that most children will need very considerable support.

■ Speech

Speech tends to develop late and at first children typically use vocal noises, gestures and sometimes signing or an assisted communication device to communicate their needs and wishes. Speech, sometimes in short phrases, may emerge, but this is not possible for all.

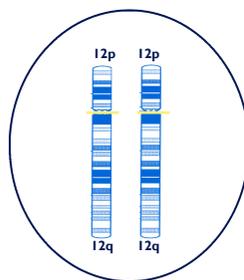
■ Behaviour

As far as we know, most children are very placid. Some are sociable with a pleasant and equable temperament but others prefer to be left alone and may dislike being handled. Any behaviour problems tend to arise in older children, especially those with greater communication and cognitive abilities.

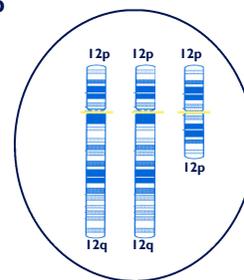
■ Sitting and moving

Babies usually roll over, sit and start moving late. The range of delay is very broad, with the age at which children take their first steps varying between two and six years. Typically, babies and young children are floppy to handle and some need supportive footwear, splints, standers and walking aids. All the same, even late sitters and walkers and those who do not walk can take part in a variety of sports activities.

A normal cell line with two chromosome 12s



A cell line with an additional isochromosome 12p



Medical concerns

■ Diaphragmatic hernia

If there is a hole in the curved muscle that separates the contents of the abdomen (tummy) from the chest, part of the intestine (gut) may develop in the chest, leaving too little room for one or both lungs to grow. Early surgical correction and replacing the intestine within the abdomen will allow the squashed lung or lungs to grow although full size may never be reached.

■ Seizures

Seizures are quite common and may start in babyhood, in childhood or not until puberty. They can usually be well controlled with medication.

■ Heart

All babies will have a thorough cardiac investigation. Around a quarter of babies with PKS are born with a heart condition, most commonly a ventricular septal defect (a hole in the wall between the two pumping chambers of the heart) or an atrial septal defect (a hole in the muscular wall between the two filling parts of the heart). Some holes will close naturally in time, but larger ones may need surgery.

■ Genitals

Slight abnormalities of the genitals are fairly common. In girls the anus and vagina may be unusually close, while in boys the genitals may be very small and the testes may not have descended into the scrotum. An operation to bring them down and anchor them may be recommended.

■ Hearing

Hearing loss is common. Some babies have the common temporary hearing loss known as glue ear that can be relieved by inserting tubes into the ear drums. Many children have a more permanent hearing loss and should be helped by wearing hearing aids.

■ Eyesight

Babies with PKS will have a thorough vision test as vision disorders are common.