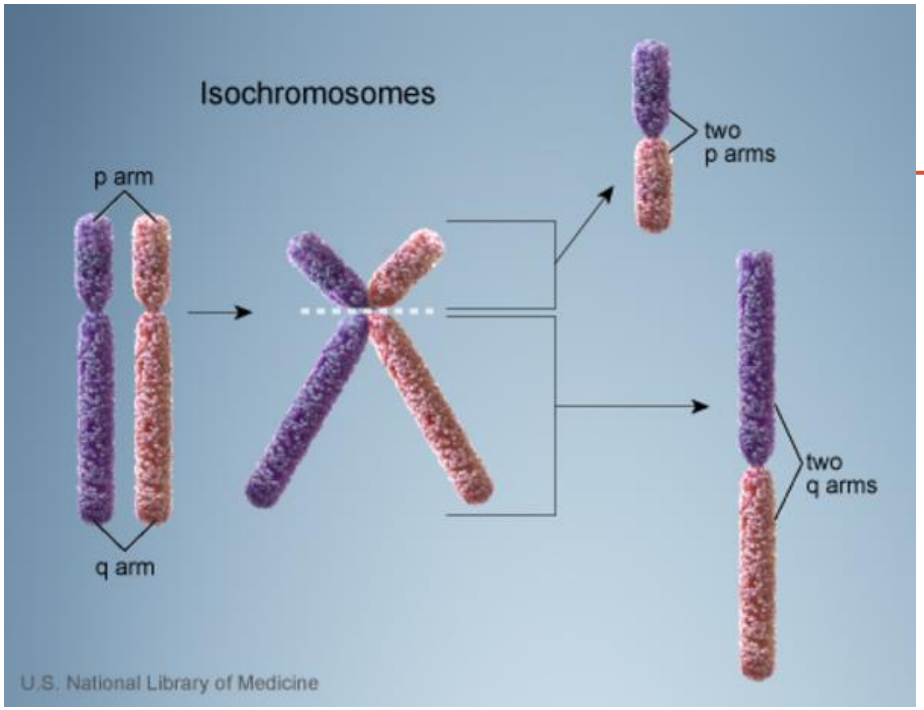
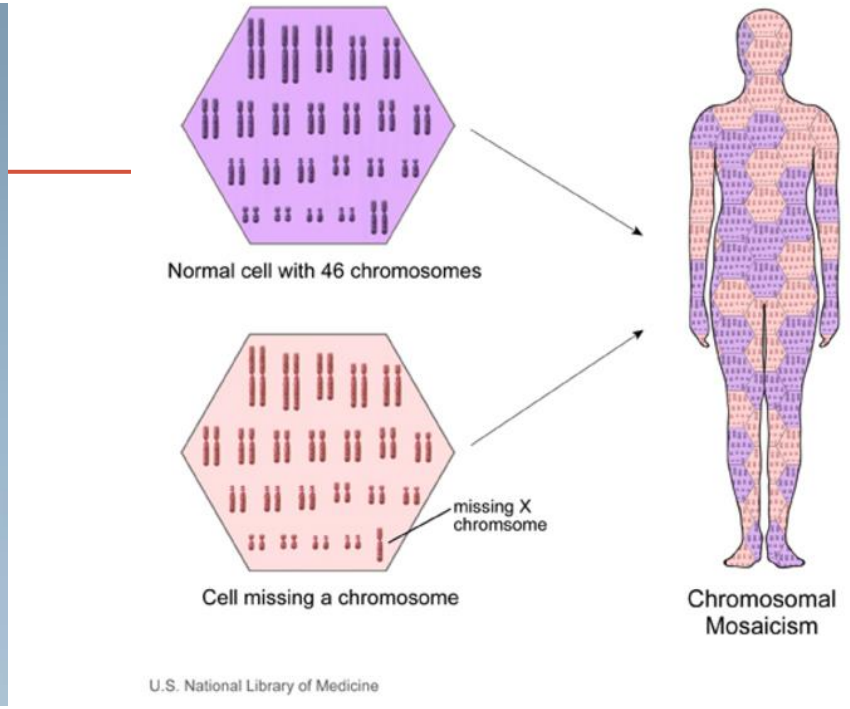
PALLISTER KILLIAN MOSAIC SYNDROME





By: Ceyda Alabacak

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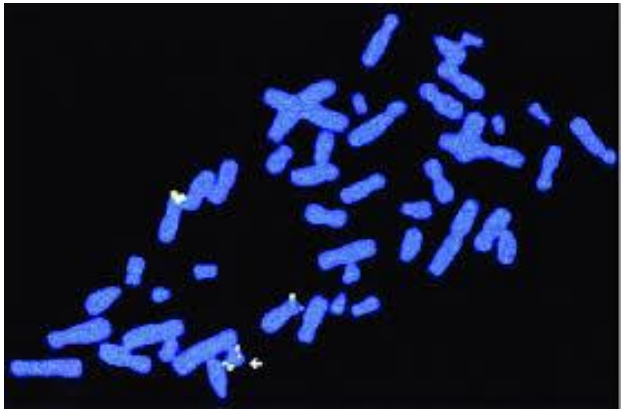
SYNONYMS OF PALLISTER KILLIAN MOSAIC SYNDROME

- Chromosome 12, Isochromosome 12p syndrome
- Killian syndrome
- Killian Teschler-Nicola syndrome
- Pallister mosaic syndrome
- Teschler-Nicola Killian syndrome
- PKS
- Isochromosome 12p syndrome
- Tetrasomy 12p, mosaic

DESCRIPTION

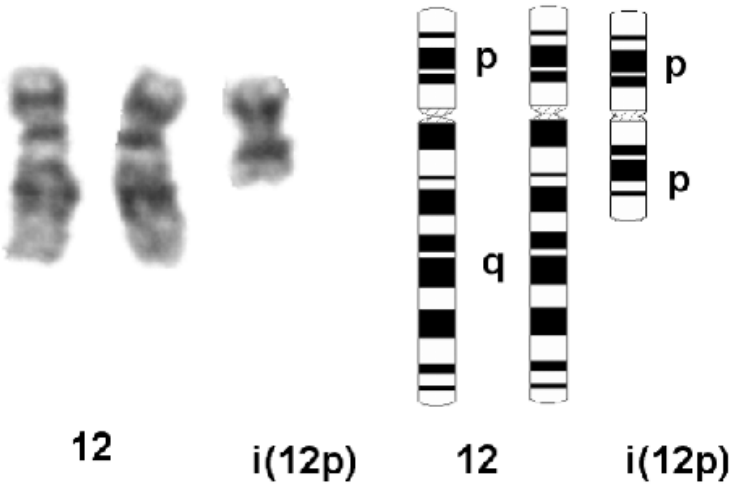
- **A rare chromosomal abnormality usually caused by the presence of an extra chromosome 12 called isochromosome 12p.**



Helpful information

Isochromosome: **A chromosome with two identical arms. Normal chromosomes have one long (q) arm and one short (p) arm, but isochromosomes have either two q arms or two p arms.**

Isochromosome 12p: **A version of chromosome 12 made up of two p arms.**



DO NOT FORGET!

- **Although Pallister-Killian Mosaic Syndrome is usually caused by an isochromosome 12p, other, more complex chromosomal changes involving chromosome 12 are responsible for the disorder in rare cases.**

SYMPTOMS

- **Hypotania*: extremely weak muscle tone**
- **Intellectual disability**

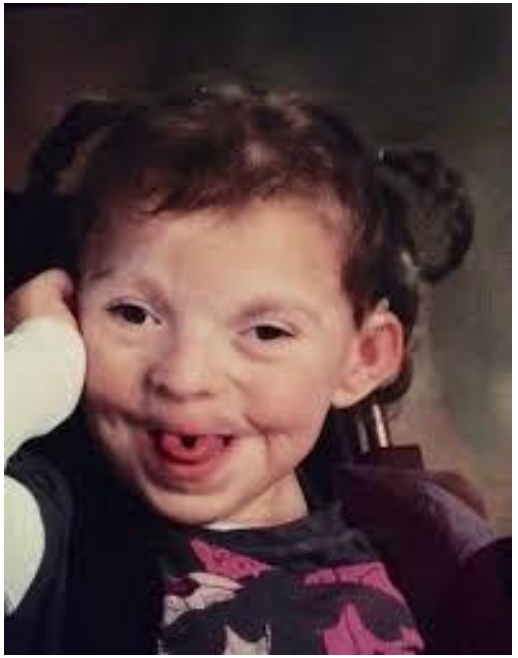
- **Breathing stoppage (temporary) or breathing trouble after birth, requiring immediate medical assistance.**

- **Distinctive facial features(coarse):**

a high, rounded forehead; a broad nasal bridge; a short nose; widely spaced eyes; low-set ears; rounded cheeks; and a wide mouth with a thin upper lip and a large tongue.

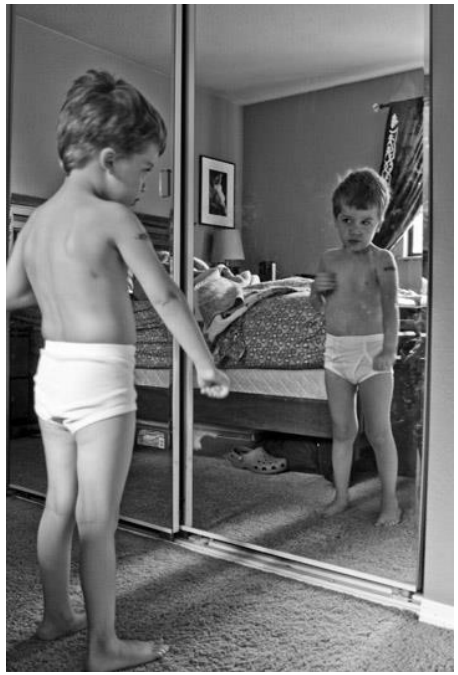
- **Sparse hair**

- **Areas of unusual skin coloring (pigmentation) other birth defects.**



HYPOTANIA*

- Hypotonia causes difficulty breathing and problems with feeding.
- Hypotonia also interferes with the normal development of motor skills such as sitting, standing, and walking.
- About 30 percent of affected individuals are ultimately able to walk without assistance.
- Additional developmental delays result from intellectual disability, which is usually severe to profound.
- Speech is often limited or absent in people with this condition.

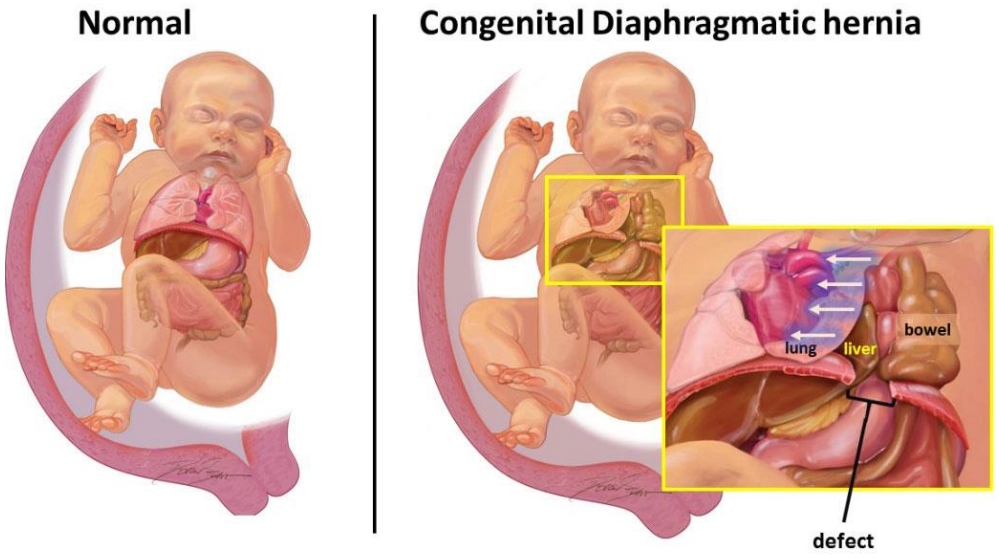


ADDITIONAL SYMPTOMS OF PALLISTER KILLIAN MOSAIC SYNDROME

- **Hearing loss**
- **Vision impairment**
- **Seizures**
- **Extra nipples**
- **Genital abnormalities**
- **Heart defects**
- **Skeletal abnormalities (extra fingers and/or toes, Large big toes halluces, unusually short arms and legs.)**
- **Congenital diaphragmatic hernia****

Congenital diaphragmatic hernia:

- A hole in the muscle that separates the abdomen from the chest cavity (the diaphragm). This potentially serious birth defect allows the stomach and intestines to move into the chest, where they can crowd the developing heart and lungs.



TREATMENT AND MANAGEMENT OF PALLISTER KILLIAN MOSAIC SYNDROME

- **Use of breathing aids (immediately after birth), in case of severe respiratory condition.**
- **Diaphragmatic hernia may require prompt and early surgical intervention.**
- **Use of hearing and vision aids to help with poor hearing and sight.**

TREATMENT AND MANAGEMENT OF PALLISTER KILLIAN MOSAIC SYNDROME

- **Correctional surgery is performed to rectify hand and feet abnormalities (such as additional fingers or toes, fused skin between the fingers and toes)**
- **Epileptic seizures are managed symptomatically with suitable medications.**
- **If there are developmental delays, then suitable supportive care and therapies are administered.**
- **Surgical treatment of heart defects.**

TREATMENT AND MANAGEMENT OF PALLISTER KILLIAN MOSAIC SYNDROME

- **Young children should undergo regular (annual) medical examination/screening, in order for the healthcare provider to assess their growth, and to also monitor for any abnormal developmental conditions**
- **Surgical procedures and braces are used to treat and rectify curvature of the spine**
- **Surgical treatment of heart defects**

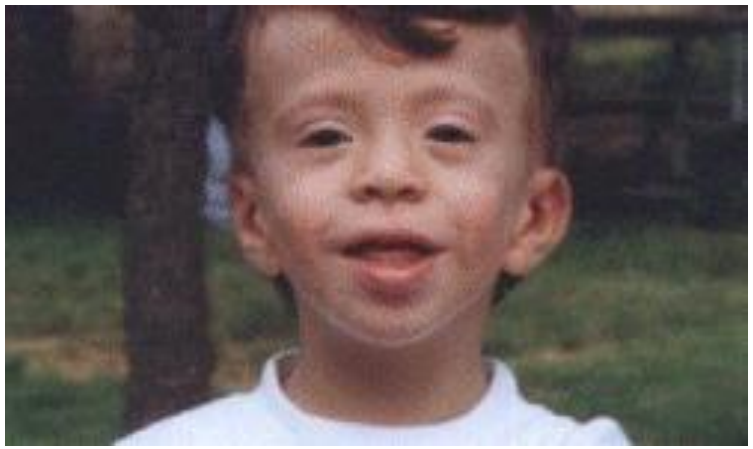
PKS KIDS IS ABOUT...

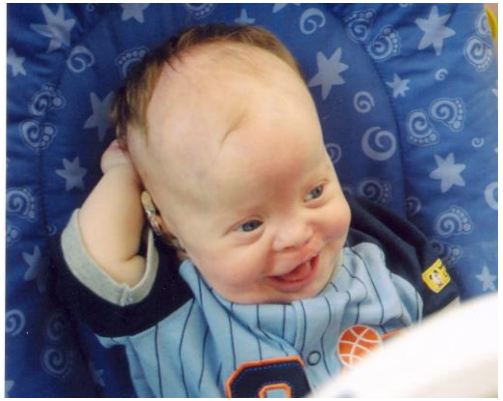
Kids

PKS Kids knows the bottom line is our children. These angels need our help and our love. In the end, everything PKS Kids does is done to give hope and help to the families who need it. To this end, we offer Giving Grants to families who need assistance with medical and therapeutic bills or equipment.

THE DISORDER VARIES

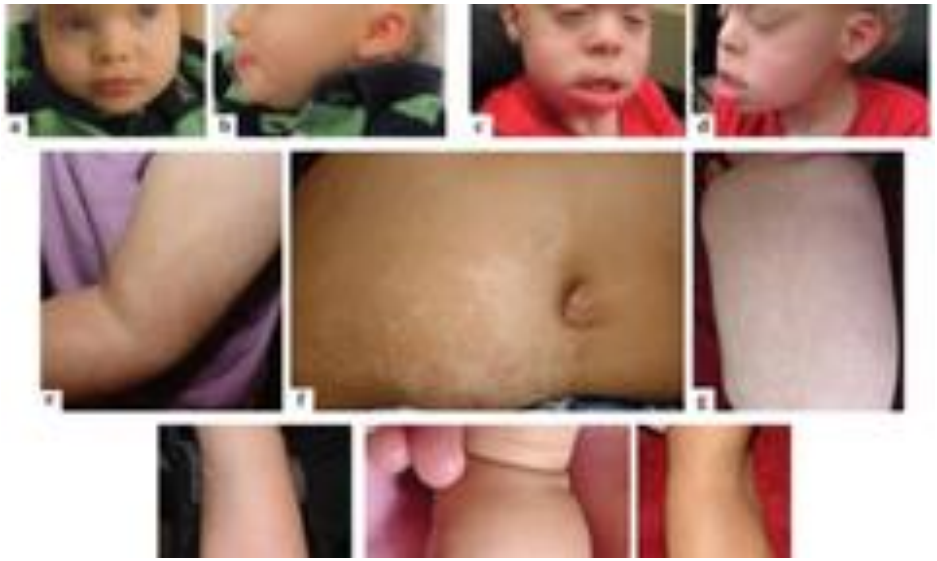
- The **signs and symptoms** of Pallister-Killian mosaic syndrome **vary.**
- For reasons that are not well understood, the severity of the effects of having mosaic tetrasomy 12p varies enormously between people.
- Therefore**, not everyone** diagnosed as having mosaic tetrasomy 12p will have classical Pallister-Killian Syndrome. **Only those who are recognisably affected by the extra DNA** are described as having classical PKS

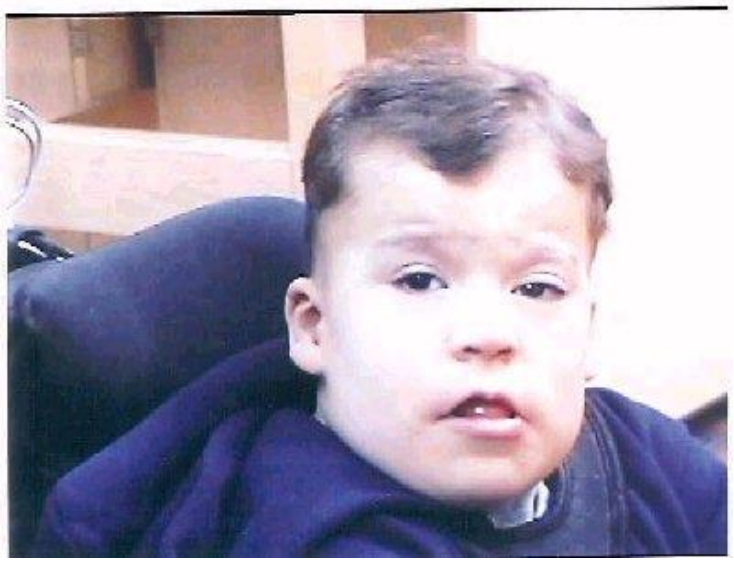




- The most severe cases involve birth defects that are life-threatening in early infancy**. However, several affected people have had milder features,** including mild intellectual disability and less noticeable physical abnormalities.

- Treatment depends upon the specific symptoms present in **each individual.** Treating medical and developmental problems early can help to optimize outcome.





GENES ON CHROMOSOME 12

- **Chromosome 12 likely contains 1,100 to 1,200 genes that provide instructions for making proteins. These proteins perform a variety of different roles in the body.**

PARTIAL LIST OF GENES ON HUMAN CHROMOSOME 12

- **ACAD10: encoding protein Acyl-CoA dehydrogenase family, member 10**
- **ACSS3: encoding protein Acyl-CoA synthetase short-chain family member 3**
- **ACVRL1: activin A receptor type II-like 1f**
- **APOF: encoding protein Apolipoprotein F**
- **APOLD1: apolipoprotein L domain containing 1**
- **ARL6IP4: encoding protein ADP-ribosylation-like factor 6 interacting protein 4**
- **ARPC3: encoding protein Actin-related protein 2/3 complex subunit 3**
- **Asun: encoding protein Protein asunder homolog (Asun)**
- **ATG101: Autophagy-related protein 101**
- **BCAT1: encoding protein Branched chain amino acid transaminase 1**
- **C12orf42: encoding protein uncharacterised chromosome 12 open reading frame 42**
- **C12orf43: encoding protein. Uncharacterized.**
- **C12orf60: encoding protein Uncharacterized protein C12orf60**

HOW THE ANOMALY PRODUCES SYMPTOMS

- **The disorder manifests in the womb, before birth of the child. Hence, it is difficult to predict the outcome of the condition.**

- **Growth from childhood to teenage, and into adulthood, gradually reveals the extent of the individual's specific condition and severity of their signs and symptoms.**

▼ HISTORY

- **The syndrome was independently reported by Philip Pallister et al. (1977) and Maria Teschler-Nicola and Wolfgang Killian (1981) because of the characteristic combination of clinical manifestations, especially the combination of coarse face, pigmentary skin anomalies, localized alopecia, profound mental retardation and seizures, and the relatively frequent occurrence of diaphragmatic defects and supernumerary nipples.**

Dr. Pallister with Aidan
PKS FACE2FACE Conference 2006

Maria Teschler-Nicola

▼ HISTORY

- **The presence of tetrasomy 12p was not recognized because chromosomal studies of fibroblasts were not done.**

- **According to Pallister (2003), chromosome studies of fibroblasts were performed by his colleague Lorraine Meisner, but the anonymous chromosome was misinterpreted.**

Lorraine Faxon Meisner

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