Research and Medical

Names it has been called:

-Pallister-Killian Syndrome (PKS)
-Pallister Mosaic Syndrome
-Killian/Teschler-Nicola Syndrome
-Tetrasomy 12p
-Mosaic isochromosome 12p syndrome

Current research is being done by geneticist, Dr. Ian Krantz at Children's Hospital of Philadelphia.

Dr. Francis Filloux, pediatric neurologist, of the University of Utah has taken a special interest in the workings of seizures and neurology in those with Pallister-Killian Syndrome and to that end is collecting data from PKS families.

Since 2006, we have hosted a biennial medical conference for families, caregivers and interested medical personnel. Please contact us at info@pkskids.net if you'd like more information.

PKS Kids
PO Box 12211
Green Bay, WI 54307

info@pkskids.com
www.pkskids.com

What is PKS?

Pallister-Killian mosaic syndrome is typically caused by the presence of an two copies of the short arm of chromosome 12 (12p). Children with this syndrome suffer low muscle tone, seizures, diaphragmatic hernias, global developmental delays, mental retardation, hearing and vision loss among other traits.

How do you recognize PKS?

Visual characteristics include: a full lower lip, flat nasal bridge, high forehead, extra nipples, short fingers, high arched palate, hypopigmentation, and a unique hair pattern early in life. Children with PKS look very much alike!

Diagnostic testing should include a skin biopsy, buccal smear or an array blood test.

Are you a medical professional with questions?

Please contact Alisha Wilkens, Genetic Counselor at The Children's Hospital of Philadelphia. 215-590-2920 or wilkens@email.chop.edu

Visiting our website will help you see the unique physical characteristics those with PKS have. On our medical page, you'll find more information and resources as well as stories of many children with PKS.

www.pkskids.com
info@pkskids.net

PALLISTER-KILLIAN SYNDROME

Medical Brochure

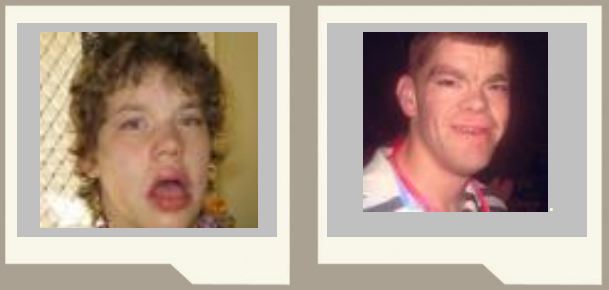
Education, Awareness, and Support for those touched by Pallister-Killian Syndrome.

www.pkskids.com

PALLISTER-KILLIAN SYNDROME

EDUCATION ~ AWARENESS ~ SUPPORT

www.pkskids.com



Our mission is to promote research, provide education and raise awareness within the medical community in order to ensure early diagnoses of children with Pallister-Killian Syndrome (PKS). To provide resources and support to families, therapists and caregivers of children with PKS.

Visit www.pkskids.com
www.pkskids.ning.com

Extremities:

- broad hands with short fingers
- shortening of arms and legs
- 5th finger clinodactyly (in-turning)
- distal digital hypoplasia
- postaxia/preaxial polydactyly (extra fingers and toes)
- single palmar crease

Ophthalmologic:

- hypertelorism, narrow palpebral fissures, epicanthi, ptosis
- sparse eyebrows, nystagmus, optic nerve atrophy, amblyopia, fundus hypopigmentation

GI:

- intestinal malrotation
- umbilical and inguinal hernias/ omphalocele
- imperforate anus, anal
- diaphragmatic hernias
- atresia.stenosis

GU:

- renal anomalies
- hypospadias

Growth:

- normal/increased length, weight and head circumference at birth
- postnatal growth deceleration of length and head circumference
- obesity

Neuro/Developmental:

- cognitive deficits range from mild to profound mental retardation
- seizures (epilepsy) (at least 40%)-
- structural brain differences (cortical atrophy, ventricular dilatation, dydrocephalus, thickened cortex, micropolygyria, heterotopia, agenesis of corpus callosum, pineal gland tumor)
- hearing loss and vision impairment
- severe psychomotor impairment

Craniofacial:

- sparse anterior scalp hair (temporal areas) especially in infancy
- sparse eyebrows and eyelashes
- coarse facial features
- prominent high forehead
- upslant to palpebral fissures
- hypertelorism (widely spaced eyes)
- strabismus
- epicanthal folds
- retinal pigmentary changes
- flat, broad nasal root with short anteverted (upturned) nose
- chubby cheeks
- long philtrum, thin upper lip with "cupid's bow" shape
- protruding lower lip, macrostomia (large mouth)
- cleft palate, bifid uvula, small chin (micrognathia)
- delayed tooth eruption
- large ears with thick lobes/helical ear pits
- midface malformations
- stenosis of external auditory canals
- short neck
- macroglossia/prognathism in older children

Skin:

- streaks of hyper/hypopigmentation

Cardiac:

- congenital heart defect (~25%) (VSD), Coarc, AS, ASD, ToF, HCM,HLH
- pericardial agenesis, lymphedema

Other:

- -accessory nipples
- -hypermobile joints
- -kyphoscoliosis
- -hemihypertrophy
- -congenital hip dislocation and dysplasia