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What is The Phelan-McDermid Syndrome?

BY CHERYL WOOD

22q13 Deletion Foundation
Phelan-McDermid Syndrome

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"Don't worry," my then first grade daughter re-assures her friend. "You can't catch it." Together they stare at her two year old brother as he scoots down the stairs on his diapered butt towards them squealing with glee. His ears are over-large, his chin a bit pointed, and his smile raises his pale puffy cheeks so that he resembles a cherub cloaked in joy.

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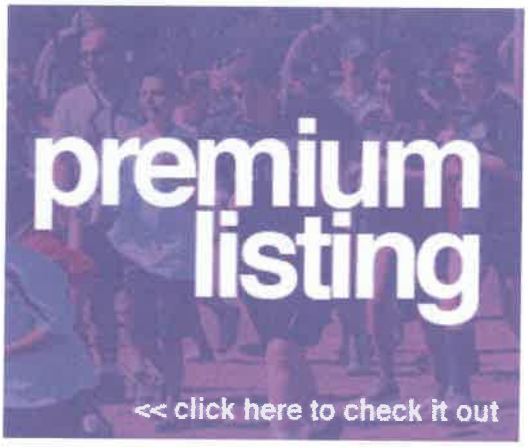
What is The Phelan-McDermid Syndrome



The Phelan-McDermid Syndrome, is a genetic condition which meant a piece on chromosome 22, long arm (q), region 1, band three, sub-band three did not exist or had been compromised. Our child was the thirty-fourth in the world to receive this diagnosis in 1998. Today there are about four hundred individuals in the world with this chromosome deletion. The condition is named for Dr. Katy Phelan and Dr.

Heather McDermid who first identified the condition. Dr. Katy Phelan brought the original first fifteen families together into what is now The Phelan-McDermid Syndrome Foundation. In 2008, over 100 families attended the support group's bi-annual conference on the East Coast.

Diagnosing The Phelan-McDermid Syndrome:

The chromosome deletion is often difficult

to see by routine chromosome studies, therefore, a special test called **FISH (fluorescence in situ hybridization)** is often used. In FISH studies, a DNA probe that is specific for the tip of the long arm of Chromosome 22 is used to see if the 22q13 segment is present or absent. If your child has Phelan-McDermid Syndrome, the 22q13 region will be present on the normal chromosome but absent on the deleted

chromosome. **A more recently developed technique, micro-array comparative genomic hybridization (CGH), is also used to identify the loss or gain of chromosome material. It is more sensitive than FISH in detecting loss of very small regions of the chromosome and can examine all of the chromosomes at once, rather than targeting only chromosome 22.** The deletion can also be

detected in cells from skin or in cells obtained for prenatal diagnostic testing.

Most Common Characteristics

Some of the most common characteristics include: Ptosis (droopy eyelid), a pointed chin, prominent poorly formed ears, puffy hands,thin flaky toenails, wide nasal bridge, hypotonia (poor muscle tone), tendency to oveheat, and lack of perspiration. Behavioral characteristics

include chewing on non food items (clothing, bedding, toys) – teeth grinding, tongue thrusting, hair pulling, aversion to clothes, decreased perception of pain and anxiety in social situations. Many children fall somewhere in the autism spectrum. Some medical issues include seizures, chronic diarrhea, macrocephaly, reactive airway disease (asthma). Very few of the children have speech or

toilet training skills.

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Phelan-McDermid Syndrome

What is? Image

Cheryl Wood

Cheryl Wood with her son Davis, her muse in life.

Cheryl holds an M.B.A. from UC Irvine, and is a Broker in a ski resort, Mammoth Lakes, in the High Sierra. Her creative outlets include

writing and designing art quilts. Periodically she will pursue various fitness ventures, the last of which was a marathon in December 2008. She likens the experience of her first marathon to raising a special needs child: one foot in front of the other until they tell you to stop. If you smile, the group running alongside you smiles back. If you fatigue, fake it 'til you make it. Cheryl serves as a Director on the Board of Directors for The Mammoth Lakes Foundation which supports higher

education and the arts. From 1998 to 2002 she created and co-created various writing programs for locals and visitors to enjoy honing their craft in a natural outdoor setting. Her current focus is her manuscript called The Thirty-Third Child (in the world) which is intended to inspire one for the journey along a life less traveled.

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