DISABILITY AWARENESS: POTOCKI-LUPSKI SYNDROME

by Baylor College of Medicine Staff

What is Potocki-Lupski Syndrome (PTLS)?
Dup 17p11.2 syndrome is a rare genetic disorder in which individuals carry an extra copy of a tiny portion of chromosome 17.

What causes PTLS?
The duplication on chromosome 17p is caused by an abnormality that occurs during the formation of the egg or sperm. There is nothing that a parent might have done to cause this condition, nor is there anything that a parent could have done to prevent the abnormality from occurring.

What should I expect in the future for my child with PTLS?
Every individual with PTLS is unique and it is impossible to know exactly what to expect. However, most individuals will have some degree of developmental delay, primarily speech delay. In addition, many individuals display some behaviors similar to those seen in patients with autism spectrum disorders.

Does this mean that my child with PTLS will have autism?
No, but they might show some features such as difficulty with interpersonal skills, repetitive behaviors, or other unusual behaviors. Only a detailed psychological evaluation can determine whether your child has features that are consistent with the diagnosis of autism or an autism-spectrum disorder.

Is there a cure for PTLS?
Because the duplication of genetic material exists in every cell of the body, there is no way to “cure” the syndrome at present. However, physical / occupational / speech therapy can help children overcome some of the developmental delays. As more patients are evaluated with PTLS, the hope is that we will develop specific educational interventions to improve communication skills in persons with PTLS. If significant cardiac defects or sleep apnea are detected, these can often be treated medically or surgically.

Are other family members at risk for having a child with PTLS?
This question can only be answered by your geneticist or genetic counselor. In most cases, PTLS occurs only in the affected child and there is a very low risk of recurrence in the family. However, in a small fraction of cases an otherwise unaffected parent can be found to have a rearrangement of the 17p11.2 region. In this case, the recurrence risk would be higher and the parents should receive detailed genetic counseling. As with many genetic syndromes, we recommend for parents to be tested for their child’s duplication in order to provide the most accurate recurrence risk counseling.

Where can I get more information about PTLS?
Since this syndrome has only recently been described, physicians are still in the process of learning about it. If you would like to share information with your primary physician, we will provide you with a reprint of our recently published article upon request. Joining the family support group is also a great way to share information and ideas.

How can I help?
We have established a clinical research protocol and a molecular research protocol at Texas Children’s Hospital in order to learn more about dup 17p11.2 syndrome. Please feel free to contact our study coordinator for more details.

http://www.bcm.edu/genetics/potocki_lupski/index.cfm?PMID=10628