SickKids | Genome Diagnostics

Trismus-Pseudocamptodactyly Syndrome

Trismus-pseudocamptodactyly syndrome is a disorder caused by a deficiency of a perinatal skeletal myosin heavy chain. Patients with trismus pseudocamptodactyly syndrome may show cardiac myxomas, spotty skin pigmentation alone or in combination with cutaneous lesions. Most affected individuals have distal arthrogryposis, including pseudocamptodactyly of the hands and feet, trismus, or both, which improves symptomatically with aging.

Genetics

Trismus-pseudocamptodactyly syndrome is an autosomal dominant disorder caused by mutations in the *MYH8* gene, located on chromosome 17 (17p13.1). A single mutation, p.Arg674Gln, has been described in several families of Belgian descent.

Trismus-pseudocamptodactyly syndrome is present when an individual has one copy of the defective *MYH8* gene. Affected individuals have a 50% chance of transmitting the disorder to each child. There is a 50% chance that the affected individual's offspring will not be affected with Trismus pseudocamptodactyly syndrome.

Who Should be Tested?

- Individuals clinically suspected of being affected with Trismus pseudocamptodactyly syndrome
- Individuals with a family history of Trismus pseudocamptodactyly syndrome
- Pregnancies at risk of being affected with Trismus pseudocamptodactyly syndrome

Test Methods

- Complete sequencing of the 38 exon coding region and flanking exon/intron boundaries of the *MYH8* gene to identify point mutations.

Potential Outcomes & Interpretation of Test Results

***MYH8* Gene Mutations**	**Explanation**
None detected	This result does not support a diagnosis of Trismus-pseudocamptodactyly syndrome
Mutation detected	This result supports a diagnosis of Trismus-pseudocamptodactyly syndrome

For More Information

Online Mendelian Inheritance in Man http://www.ncbi.nlm.nih.gov/omim/ Item # 158300

To locate a genetics center near you, please visit the Canadian Association of Genetic Counsellors website at www.cagc-accg.ca or the National Society of Genetic Counsellors website at www.nsgc.org

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1. Current molecular testing may not detect all possible mutations in this gene. A negative test result does not rule out the possibility that the individual carries a rare MYH8 mutation not detected by this assay.

2. Test results should be interpreted in the context of clinical findings, family history and other laboratory data.

3. This test was developed and its performance characteristics validated by the Genome Diagnostics Laboratory at the Hospital for Sick Children. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes.