MSMO1-related Psoriasiform Dermatitis: MSMO1 Gene Sequencing

Test Code: SMSMO
Turnaround time: 6 weeks
CPT Codes: 81479 x1

Condition Description

He et al. reports a female with severe ichthyosiform erythroderma. It affects her entire body with the exception of her palms. Symptoms were first noted at 2 years of age and by age 6, had progressed to involve the remainder of her body. Additional clinical features included congenital cataracts, mild developmental delay, microcephaly, and failure to thrive. Mutation analysis of the MSMO1 gene (4q32-q34), also known as the SC4MOL gene, identified two different mutations. One was subsequently found in the patient’s father and the other was found in the patient’s mother demonstrating autosomal recessive inheritance. Neither mutation was identified in 2876 alleles from population controls. Deficiency of sterol-C4-methyl oxidase represents a biochemical defect in the cholesterol synthesis pathway for which the clinical spectrum remains to be defined.

References:

- OMIM #607545: SC4MOL gene

Genes

MSMO1, SC4MOL

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of MSMO1-related psoriasiform dermatitis.
- Carrier testing in adults with a family history of MSMO1-related psoriasiform dermatitis.

Methodology

PCR amplification of 5 exons contained in the MSMO1 gene is performed on the patient’s genomic DNA. Direct sequencing of amplification products is performed in both forward and reverse directions, using automated fluorescence dideoxy sequencing methods. The patient’s gene sequences are then compared to a normal reference sequence. Sequence variations are classified as mutations, benign variants unrelated to disease, or variations of unknown clinical significance. Variants of unknown clinical significance may require further studies of the patient and/or family members. This assay does not interrogate the promoter region, deep intronic regions, or other regulatory elements, and does not detect large deletions.

Detection

Clinical Sensitivity: Unknown. Mutations in the promoter region, some mutations in the introns and other regulatory element mutations cannot be detected by this analysis. Large deletions will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's clinical and/or biochemical phenotype.

Analytical Sensitivity: ~99%

Specimen Requirements

Submit only 1 of the following specimen types

* Preferred specimen type: Whole Blood

Type: Whole Blood

Specimen Requirements:

In EDTA (purple top) or ACD (yellow top) tube:
Infants (2 years): 3-5 ml
Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

Type: Saliva

Specimen Requirements:

Oragene™ Saliva Collection kit (available through EGL) used according to manufacturer instructions.
Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Related Tests**

- Custom diagnostic mutation analysis (KM) is available to family members if mutations are identified by targeted mutation testing or sequencing analysis.
- Prenatal testing is available only for known familial mutations to individuals who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.