### 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**

#### Type: Saliva

**Specimen Requirements:**  
Oragene™ Saliva Collection Kit  
Oragene™ Saliva Collection Kit used according to manufacturer instructions. Please contact EGL for a Saliva Collection Kit for patients that cannot provide a blood sample.

**Specimen Collection and Shipping:**  
Please do not refrigerate or freeze saliva sample. Please store and ship at room temperature.

#### Type: Whole Blood (EDTA)

**Specimen Requirements:**  
EDTA (Purple Top)  
Infants and Young Children (2 years of age to 10 years old): 3-5 ml  
Older Children & Adults: 5-10 ml  
Autopsy: 2-3 ml unclotted cord or cardiac blood

**Specimen Collection and Shipping:**
Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

**Type:** DNA, Isolated

**Specimen Requirements:**
- Microtainer
- 8µg

Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

**Specimen Collection and Shipping:**
Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample 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.