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**Lab Dept:** Anatomic Pathology

**Test Name:** SOTOS SYNDROME (NSD1)  
DELETION/DUPLICATION

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***General Information***

**Lab Order Codes:** SOTD

**Synonyms:** NSD1 Deletion Duplication

**CPT Codes:** 81405 – NSD1 (nuclear receptor binding SET domain protein 1), deletion/duplicaton – Molecular Pathology, Level 6

**Test Includes:** Deletion/Duplication analysis of the NSD1 gene by MLPA to identify deletions/duplications of one or more exons.

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***Logistics***

**Test Indications:** Sotos syndrome is characterized by characteristic facial features, developmental delay, and increased height and head circumference. Other features may include: neonatal jaundice, scoliosis, seizures, strabismus, conductive hearing loss, congenital heart defects, renal abnormalities, and behavior problems.

Microdeletions and mutations of the NSD1 gene have been identified in approximately 80% of patients with a clinical diagnosis of Sotos syndrome.

**Lab Testing Sections:** Anatomic Pathology - Sendouts

**Referred to:** University of Chicago Genetic Services Laboratory (UCHI Test: 1223)

**Phone Numbers:** MIN Lab: 612-813-6280

STP Lab: 651-220-6550

**Test Availability:** Daily, 24 hours

**Turnaround Time:** 4 weeks

**Special Instructions:** A completed University of Chicago Requisition and Consent forms must accompany the specimen or patient to the laboratory. Forms can be found at the following addresses.

Requisition:

[http://dnatesting.uchicago.edu/sites/default/files/03ReqForm\\_2.pdf](http://dnatesting.uchicago.edu/sites/default/files/03ReqForm_2.pdf)

Consent:

<http://dnatesting.uchicago.edu/sites/default/files/05ConsentForm.pdf>

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***Specimen***

<b>Specimen Type:</b>	Whole blood
<b>Container:</b>	Lavender top (EDTA) tube
<b>Draw Volume:</b>	10 mL (Minimum: 3 mL) blood
<b>Processed Volume:</b>	Same as Draw Volume
<b>Collection:</b>	Routine venipuncture
<b>Special Processing:</b>	Lab Staff: Do Not centrifuge. Specimen should remain in original collection container. Specimen should remain at room temperature. Send Monday-Thursday overnight. If specimens are collected on Friday, Saturday or Sunday, specimen can be held at Children's refrigerated and shipped for appropriate delivery.
<b>Patient Preparation:</b>	None
<b>Sample Rejection:</b>	Frozen specimens, mislabeled or unlabeled specimens

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***Interpretive***

<b>Reference Range:</b>	Interpretive report
<b>Critical Values:</b>	N/A
<b>Limitations:</b>	N/A
<b>Methodology:</b>	MLPA
<b>References:</b>	<a href="#">University of Chicago Genetic Services Laboratory</a> March 2018 Phone: 1-888-824-3637 Fax: 773-702-9130
<b>Updates:</b>	2/11/2013: CPT update