
Lab Dept: **Anatomic Pathology**

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

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.

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

Consent:

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

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube
Draw Volume:	10 mL (Minimum: 3 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture
Special Processing:	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.
Patient Preparation:	None
Sample Rejection:	Frozen specimens, mislabeled or unlabeled specimens

Interpretive

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