
Lab Dept: **Anatomic Pathology**

Test Name: **SOTOS SYNDROME (NSD1) GENE SEQUENCING**

General Information

Lab Order Codes: SOTS

Synonyms: NSD1 Gene Sequencing

CPT Codes: 81406 – NSD1 (nuclear receptor binding SET domain protein 1), full gene sequence – Molecular Pathology, Level 7

Test Includes: Full gene sequencing of the entire coding region for the NSD1 gene.

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.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: University of Chicago Genetic Services Laboratory (UCHI Test: 1222)

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 blood collection
Special Processing:	Lab Staff: Do Not centrifuge. Specimen should remain in original collection container. Specimen should remain at room temperature. Send Sunday-Thursday overnight. If specimens are collected on Friday or Saturday, 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:	None
Methodology:	Gene sequencing
References:	University of Chicago Genetic Services Laboratory March 2018
Updates:	1/9/2013: 2013 CPT update