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 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