ARUP LABORATORIES

A nonprofit enterprise of the University of Utah and its Department of Pathology

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THIS IS NOT A TEST REQUEST FORM. Please complete and submit with the test request form or electronic packing list.

SMITH-LEMLI-OPITZ SYNDROME (*DHCR7*) TESTING PATIENT HISTORY FORM

Patient Name: _____ **Date of Birth:** _____ **Sex:** ☐ Female ☐ Male

Ordering Provider: _____ **Provider's Phone:** _____

Practice Specialty: _____ **Provider's Fax:** _____

Genetic Counselor: _____ **Counselor Phone:** _____

Patient's Ethnicity/Ancestry (check all that apply)

☐ African American/Black ☐ Asian ☐ Hispanic ☐ White ☐ Other: _____

List country of origin (if known): _____

Does the patient have symptoms? ..... ☐ No ☐ Yes (check all that apply and describe)

☐ Cardiac defect(s); describe: _____

☐ Characteristic facial features
☐ Cleft palate
☐ Congenital cataracts
☐ Developmental delay/intellectual disability:
  ☐ Mild
  ☐ Moderate
  ☐ Severe
  ☐ Autistic spectrum disorder
☐ Feeding difficulty

☐ Genital anomalies:
  ☐ Ambiguous genitalia
  ☐ Hypospadias
  ☐ Cryptochidism
  ☐ Other: _____
☐ Growth deficiency:
  ☐ Prenatal
  ☐ Postnatal
  ☐ Short stature
☐ Hypotonia
☐ Microcephaly

☐ Renal anomalies; describe: _____

☐ Sensorineural hearing loss
☐ Skeletal findings:
  ☐ Postaxial polydactyly
  ☐ 2-3 toe syndactyly
☐ Ultrasound findings;
  ☐ Describe: _____
☐ Other symptom(s): _____

Laboratory Findings

Serum 7-dehydrocholesterol (7-DHC): ☐ Normal ☐ Elevated: _____ ☐ Unknown ☐ Not performed

Serum cholesterol:..... ☐ Normal ☐ Low: _____ ☐ Unknown ☐ Not performed

☐ Abnormal maternal serum screening;
values in multiples of the median (MoM): AFP: _____ hCG: _____ uE3 (estriol): _____ DIA: _____

Has the patient undergone previous DNA testing for Smith-Lemli-Opitz syndrome (SLOS)?..... ☐ No ☐ Yes ☐ Unknown

If yes, describe the test(s) and results: _____

Is there any relevant family history? ..... ☐ No ☐ Yes ☐ Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age of onset:

Has DNA testing been performed for the family member(s)? ..... ☐ No ☐ Yes ☐ Unknown

If yes, attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing).

Check the test you intend to order.

☐ **2011457 Smith-Lemli-Opitz Syndrome *(DHCR7)* Sequencing:** Sequencing of *DHCR7* coding regions and intron/exon boundaries; clinical sensitivity is approximately 96%.

☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a mutation previously Identified in a family member; a copy of relative's lab result is REQUIRED.

☐ **3003144 Deletion/Duplication Analysis by MLPS:** Tests for large deletion/duplication previously identified in a family member; a copy of a relative's lab report is REQUIRED.

Master Label

For questions, contact an ARUP genetic counselor at 800-242-2787 ext. 2141.