ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY
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 fill out this form and submit it with the test request form or electronic packing list.

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

Patient Name _____ Date of Birth _____ Sex ☐ F ☐ M

Physician _____ Physician Phone _____

Practice Specialty _____ Physician Fax _____

Genetic Counselor _____ Counselor Phone _____

Patient's Ethnicity (check all that apply)

☐ African-American ☐ Asian ☐ Hispanic ☐ Native American
☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other: _____

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 ☐ Cryptorchidism ☐ 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.

Molecular genetic testing for Smith-Lemli-Opitz syndrome:
☐ **2011457 Smith-Lemli-Opitz Syndrome (*DHCR7*) Sequencing**
☐ **2011704 Smith-Lemli-Opitz Syndrome (*DHCR7*) Sequencing, Fetal**
Targeted testing for known familial mutation (a copy of a relative's lab result is REQUIRED):
☐ **2001961 Familial Mutation, Targeted Sequencing**
☐ **2001980 Familial Mutation, Targeted Sequencing, Fetal:** Prenatal testing.

Master Label

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