ARUP LABORATORIES

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform molecular genetic testing.
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_____ /_____/_____Gender [ ] F [ ] M

Physician _____ Physician Phone (_____)_____Practice Specialty_____

Genetic Counselor _____ Counselor Phone (_____)_____

Patient's Ethnicity (check all that apply)
[ ] African American [ ] Ashkenazi Jewish [ ] Asian [ ] Caucasian
[ ] Hispanic [ ] Middle Eastern [ ] Native American [ ] Other _____

Does the patient have SYMPTOMS of Smith-Lemli-Opitz syndrome (SLOS)? [ ] No [ ] Yes; check all that apply

[ ] 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: _____

Laboratory test RESULTS:
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 SLOS? [ ] No [ ] Yes [ ] Unknown
If yes, please describe test(s) and results:_____

Does the patient have a FAMILY HISTORY of SLOS? [ ] No [ ] Yes [ ] Unknown
If yes, attach a PEDIGREE or specify the RELATIONSHIP of the family member(s) to the patient and detail the symptoms/age of onset in each symptomatic relative: _____
_____

Has DNA testing been performed for these family member(s)? [ ] No [ ] Yes [ ] Unknown
If yes, please describe test(s) and results: _____

Circle the test you intend to order OR write the test name and number below:

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 mutation (laboratory report from family member REQUIRED)	
2001961	Familial Mutation, Targeted Sequencing- targeted testing for a known familial sequence mutation
2001980	Familial Mutation, Targeted Sequencing, Fetal- prenatal targeted testing for a known familial sequence mutation

Other test not listed: _____

Master Label

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