

CLINICAL INFORMATION

Diagnosis Suspected: _____

Clinical History

(Please check the appropriate symptoms and/or attach additional clinical information or summary, if relevant.)

| | | |
|------------------------------------|--------------------------------------|-----------------------------|
| G Developmental Delay | G Neurodegeneration | G Skin Changes |
| G Hypotonia * | G Leukodystrophy | G Hepatomegaly |
| G SIDS/Near SIDS | G Spasticity | G Liver Disease |
| G Failure to Thrive | G Visual Defect | G Cardiomegaly |
| G Recurrent Vomiting | G Unusual Facies | G Hypoglycemia |
| G Mental Retardation | G Cataracts * | G Metabolic Acidosis |
| G Neuronal Migration Defect | G Deafness | G Lactic Acidosis |
| G Acute Encephalopathy | G Retinal Dysplasia | G Ketonuria |
| G Macrocephaly | G Renal Cysts/Dysplasia | G Hyperammonemia |
| G Microcephaly | G Stippled Epiphyses * | G Hyperuricemia |
| G Movement Disorder | G Adrenal Hypo/Hyperplasia | G Neurotopenia |
| G Seizures | G Similarly Affected Siblings | |

* Individuals with cataracts, stippled epiphyses, hypotonia, short stature, or growth retardation require **BOTH** Plasma Very Long Chain Fatty Acids and Red Blood Cell (RBC) Plasmalogen levels.

Other Clinical Information:

| Drug and Feeding Information | Pedigree Information |
|---|-----------------------------|
| Medications and special diets or supplements <i>(please list all)</i> | |
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| | |
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Kennedy Krieger Institute Genetics Laboratory - Clinical Mass Spectrometry Section
707 North Broadway, Baltimore, MD 21205

Director: Richard I. Kelley, M.D., Ph.D. Phone: (443) 923-2782 EIN #: 52-0607971 CLIA #: 21D0649789
Associate Director: Lisa E. Kratz, Ph.D. Fax: (443) 923-2781 JCAHO#: 6252

Patient and Report Information

| | | | | | |
|-------------------|--------------------|--------------------------|-----|-----|-----------|
| Patient Last Name | Patient First Name | MI | Sex | DOB | History # |
| Sample Date | Sample ID# | Other Sample Information | | | |

| Indicate Where to Send Bill: (Laboratory or Physician only)* | | Indicate Where to Send Report: | |
|---|-----|---------------------------------------|-----|
| Name | | Name | |
| Address | | Address | |
| City, State, Zip Code | | City, State, Zip Code | |
| Phone | Fax | Phone | Fax |

Test Requisition and Sample Information (X Test(s) Desired)

| X | Test | CPT Code | Sample Types and Amounts | Cost |
|----------|---|-----------------|--------------------------------------|-------------|
| | Organic acid gas chromatography/mass spectrometry | 83918 | Urine (5ml) or CSF (1ml) | \$300 |
| | N-acetyl-l-aspartic acid (Canavan disease) | 82543 | Urine (5ml) or CSF (1ml) | \$150 |
| | Canavan disease - prenatal diagnosis | 82543 | Amniotic Fluid (10ml) | \$400 |
| | 3-Methylglutaconic Acid | 82543 | Urine (5ml) or Plasma (1ml) | \$150 |
| | 7-Dehydrocholesterol (Smith-Lemli-Opitz Syndrome) | 82542 | Plasma (1ml) | \$150 |
| | Smith-Lemli-Opitz Syndrome - prenatal diagnosis | 82542 | Amniotic Fluid (5ml) or CVS (5-10mg) | \$400 |
| | Desmosterol | 82542 | Plasma (1 ml) | \$150 |
| | Cholest 8(9) en-3β-ol (Chondrodysplasia Punctata) | 82542 | Plasma (1ml) | \$150 |
| | Cholestanol (Cerebrotendinous xanthomatosis) | 82542 | Plasma (1ml) | \$150 |
| | Sitosterol (Sitosterolemia, Phytosterolemia) | 82542 | Plasma (1ml) | \$150 |
| | Guanidinoacetic Acid + Creatine | 82544 | Urine (5 ml) or Plasma (1ml) | \$150 |
| | Mevalonic Acid | 82543 | Urine (5 ml) | \$150 |
| | Free Fatty Acids - Total level and qualitative analysis | 82725 | Urine (5ml) or Plasma (1ml) | \$200 |

Sample Collection:

Urine samples, ideally 24-hour, should be collected without preservatives and kept frozen. For organic acid analysis, an “acute” urine specimen, if relevant, should be submitted. Prenatal diagnosis should be coordinated **in advance** with Dr. Kelley or Dr. Kratz

Shipping: Specimens should be shipped **frozen** by overnight express carrier to: Kennedy Krieger Institute
Ship to arrive on weekdays only
Clinical Mass Spectrometry Section, Room 526
707 North Broadway
Baltimore, MD 21205

***Billing:** Billing is to the referring physician or laboratory. We regret that we are unable to bill patients or insurance.

PEROXISOMAL DISEASES SECTION
KENNEDY KRIEGER INSTITUTE
707 North Broadway, Room 530
Baltimore, MD 21205
TEL: (443) 923-2788, FAX: (443) 923-2755
Email: mosera@kennedykrieger.org
Steinbergs@kennedykrieger.org

Sample for DNA BASED MUTATION ANALYSIS FOR X-LINKED ADRENOLEUKODYSTROPHY (X-ALD)

SAMPLE REQUIREMENTS:

Mondays to Thursdays only for sample collection; 10 ml EDTA blood sent at room temperature by overnight delivery to above address (mutation analysis will be done by the DNA Diagnostic Laboratory at Johns Hopkins)

PATIENT INFORMATION:

Name: _____
Date of Birth: _____
Address: _____

Relationship to proband (male with X-ALD) (attach pedigree if available): _____

| <u>TESTING REQUESTED</u> | <u>CPT Codes</u> | <u>COST</u> |
|---|---|--------------------|
| Type of testing (Choose 1) | | |
| <input type="checkbox"/> Mutation Identification, proband | 83891, 83894x3, 83898x8, 83904x16, 83912 | \$1,200 |
| <input type="checkbox"/> Carrier / Heterozygote (includes plasma very long chain fatty acid) | 83891, 83894x3, 83898, 83904x2, 83912, 82726 | \$329 |
| <input type="checkbox"/> Prenatal (by prior arrangement) | 83891, 83894x3, 83898, 83904x2, 83912, 82726, 88235 | \$700 |

BILLING INFORMATION: **EXCEPT FOR INSTITUTIONAL BILLING, PAYMENT MUST ACCOMPANY THE SAMPLE**

Credit Card: Card Type: _____ Card Number: _____
Cardholders Name: _____ Expiration Date: _____

Cardholder's Signature: _____

Check (Make check out to the Kennedy Krieger Institute, PDL)

Institutional Billing Billing Address, Phone and FAX:

Authorized by: _____

COPY OF REPORT TO:

Name: _____
Address: _____
Phone: _____ Fax: _____

COPY OF REPORT TO:

Name: _____
Address: _____
Phone: _____ Fax: _____

Kennedy Krieger Institute Genetics Laboratory - Peroxisomal Diseases Section
707 North Broadway, Baltimore, MD 21205 www.genetics.kennedykrieger.org

Director: George H. Thomas, Ph.D.
Laboratory Manager: Ann B. Moser
Technical Supervisor: Richard Jones, Ph. D.

Phone: (443) 923-2760
Fax: (443) 923-2755
Email: mosera@kennedykrieger.org

EIN #: 52-0607971
JCAHO#: 6252
CLIA#: 21D0649789

Patient and Report Information

| | | | | | |
|---|--|--------------------------|---------------------------------------|-----------------|-----------|
| Patient Last Name | Patient First Name | MI | Sex | DOB (Important) | History # |
| Sample Date | Sample ID# | Other Sample Information | | | |
| Indicate Where to Send Bill: (Patient, Laboratory or Physician)* | | | Indicate Where to Send Report: | | |
| Name | | | Name | | |
| Address | | | Address | | |
| City, State, Zip Code | | | City, State, Zip Code | | |
| Phone Fax | Patient Prepayment, check or credit card* | Phone | Fax | | |

Test Requisition and Sample Information (X Test(s) Desired)

| X | Tests using Blood and Urine | CPT Code | Sample Types and Amounts | Cost |
|---|---|----------|---|-------|
| | Plasma Very Long Chain Fatty Acids: Includes Phytanic Acid (screening test for peroxisomal disorders) | 82726 | EDTA plasma or 1-3 ml whole EDTA blood; fasting or preprandial | \$165 |
| | Red Blood Cell Plasmalogen Content (screening test for rhizomelic chondrodysplasia punctata) | 82492 | RBCs from EDTA blood (discard buffy coat) or 1.5 (bare minimum)-3 ml whole EDTA blood** | \$150 |
| | Plasma Total Lipid Fatty Acid Profile: Includes C8 to C26 saturated, monounsaturated, polyunsaturated (essential), trans and branched chain fatty acids | 82492x2 | EDTA plasma or 1-3 ml whole EDTA blood; fasting or preprandial. Sample must be received by us within 24 hours of collection | \$200 |
| | Red Blood Cell Total Lipid Fatty Acid Profile: Includes C8 to C26 saturated, monounsaturated, polyunsaturated fatty acids and plasmalogens | 82492x2 | 1.5 ml (bare minimum) -3 ml whole EDTA blood; fasting or preprandial. Sample must be received by us within 24 hours of collection | \$200 |
| | Pipecolic Acid: Plasma <u>or</u> Urine | 82543 | 10 ml cleanly collected urine <u>or</u> 1 ml EDTA plasma ** | \$250 |

Sample Shipping: All samples except urine may be shipped at room temperature by overnight express. Urine for Pipecolic Acid should be shipped on wet or dry ice overnight express.

Ship samples to arrive on weekdays only to:
Kennedy Krieger Institute
Peroxisomal Diseases Section, Room 530
707 North Broadway
Baltimore, MD 21205

*Billing: Credit Card _____ # _____ Exp. Date _____ We regret that we are unable to bill insurance.
Signature _____

** If plasma pipecolic acid and/or plasmalogen content are ordered at the same time as very long chain fatty acids, all tests may be performed on a single sample of 3 ml whole EDTA blood.

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Patient and Report Information

| | | | | | |
|-------------------|--------------------|--------------------------|-----|-----------------|-----------|
| Patient Last Name | Patient First Name | MI | Sex | DOB (Important) | History # |
| Sample Date | Sample ID# | Other Sample Information | | | |

Indicate Where to Send Bill: (Patient, Laboratory or Physician)*

Indicate Where to Send Report:

| | |
|-----------------------|--|
| Name | Name |
| Address | Address |
| City, State, Zip Code | City, State, Zip Code |
| Phone Fax | Patient payment, check or credit card* Phone Fax |

Test Requisition and Sample Information (X Test(s) Desired)

| X | Tests using Cultured Cells | CPT Code | Sample Types and Amounts | Cost |
|-----|--|---|---|-------|
| | Very Long Chain Fatty Acids (Fibroblast) | 88233 82726 | 2 mm punch sterile skin biopsy in sterile tissue culture media or two T-25 flasks of fibroblasts. | \$250 |
| | Peroxisomal Plasmalogen Synthesis (Fibroblast) | 88233 82658 | 2 mm punch sterile skin biopsy in sterile tissue culture media or two T-25 flasks of fibroblasts. | \$250 |
| | Phytanic Acid Oxidase (Fibroblast) | 88233 82658 | 2 mm punch sterile skin biopsy in sterile tissue culture media or two T-25 flasks of fibroblasts. | \$250 |
| | Catalase Distribution (Fibroblast) | 88233 82657 | 2 mm punch sterile skin biopsy in sterile tissue culture media or two T-25 flasks of fibroblasts. | \$250 |
| | Diagnostic Peroxisomal Enzyme Package (Fibroblast) Includes the 4 tests above | 88233 82658x2 82657 82726 | 2 mm punch sterile skin biopsy in sterile tissue culture media or two T-25 flasks of fibroblasts. | \$700 |
| CVS | Peroxisomal Prenatal Diagnosis (For the diagnosis of X-linked ALD, Zellweger Syndrome, neonatal ALD, and rhizomelic chondrodysplasia punctata). Please circle the sample type in the box to the left. | 88235 82726 82658 (the same for CVS or AF) | Cultured CVS (two T-25 flasks) This test requires maternal cell contamination studies to be performed by the referring laboratory. | \$500 |
| AF | | | Whole amniotic fluid (15-30ml) or cultured amniocytes (two T-25 flasks). | \$500 |

Sample Shipping:

Samples must be shipped in leak proof containers completely filled with sterile tissue culture media.

Ship samples to arrive on weekdays only to:

Kennedy Krieger Institute
Peroxisomal Diseases Section, Room 530
707 North Broadway
Baltimore, MD 21205

*Billing: Credit Card _____ # _____ Exp. Date _____ We regret that we are unable to bill insurance.

Signature