NEWBORN SCREENING PROGRAM
New York State Department of Health
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Albany, NY 12208
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E-mail: nbsinfo@health.ny.gov
Website: http://www.wadsworth.org/newborn/

NEWBORN INFORMATION

Name at birth: _____
AKA: _____

Single Birth ☐ Twin A ☐ Twin B ☐ Other _____
Mother's name: _____
Date of Birth: _____

Gender: Male ☐ Female ☐
Hospital of birth: _____
Medical Record #: _____

ADRENOLEUKODYSTROPHY REFERRAL DIAGNOSIS FORM

Please complete this form in its entirety and return it to the Newborn Screening Program as soon as possible. Your response is required, as specified in Title 10 New York Code of Rules and Regulations subpart 69-1.7c.

Note: Newborn Screening results do not constitute a diagnosis. Confirmatory testing is required.

1. Select diagnosis:

[ ] No disease
[ ] No disease, female carrier of ABCD1 mutation
[ ] Expired, No diagnosis
[ ] X-linked adrenoleukodystrophy (ALD)
[ ] Definite [ ] Possible
[ ] Zellweger spectrum disorder
[ ] Definite [ ] Possible
[ ] D-Bifunctional protein deficiency
[ ] Acyl-CoA oxidase deficiency
[ ] Peroxisomal disorder of unknown etiology, X-linked ALD ruled out
[ ] Definite [ ] Probable [ ] Possible
[ ] Other, specify _____

2. Confirmatory testing

Results are required from at least the newborn. Please include parental results if available.

DATE	TEST	Newborn's Results	Mother's Results	Father's Results	Normal Range
	C26:0/C22:0				
	C24:0/C22:0				
	C26:0				
	Plasmalogen				

3. Fibroblast Studies [ ] Normal [ ] Abnormal [ ] Not Done

If abnormal, please provide interpretation or include report: _____

4. *ABCD1* MLPA: [ ] Normal [ ] Abnormal [ ] Not Done
Please include report

5. Other Genetic Testing (i.e PEX genes): [ ] Normal [ ] Abnormal [ ] Not Done
Please include report

6. Abnormal clinical findings? Yes No

7. If yes, please specify

[ ] Hypotonia
[ ] Poor feeding
[ ] Distinctive facies
[ ] Seizures
[ ] Hepatic dysfunction and abnormal coagulation studies
[ ] Renal or liver cysts
[ ] Jaundice
[ ] Bone stippling
[ ] Retinal dystrophy
[ ] Sensorineural hearing loss
[ ] Abnormal coagulation studies
[ ] Other, Specify _____

8. Maternal Ethnicity _____ Paternal Ethnicity _____

9. Was this newborn previously known to be at increased risk for this disorder?
[ ] No [ ] Yes, family history [ ] Yes, prenatal testing [ ] Yes, preconception testing

COMMENTS: _____

PHYSICIAN'S SIGNATURE: _____DATE:_____

PRINT NAME: _____FACILITY/PRACTICE: _____