Neurogenetics DNA Diagnostic Lab
Massachusetts General Hospital
Center for Human Genetic Research
CRP Building North, Suite 5300
185 Cambridge Street
Boston, MA 02114

Lab Coordinator Phone: 617-726-5732
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Neuronal Ceroid Lipofucinoses [NCL]
Clinical Features Checklist

Date:

Patient Name:
D.O.B.:
Person completing form:
Tel #:
Fax #:

Relationship to Proband:
Email:

Clinical Features
Ethnicity:
Date of Last exam: / /

Development [note current level of function]
☐ Normal ☐ Delayed

Regression [note current level of function]
☐ Yes ☐ No Age at which regression began: ________________________

Current Function:	**Normal**	**Abnl**	**Regression?** ***If yes, please note age of onset***
Fine motor:	☐	☐	__________________
Gross motor:	☐	☐	__________________
Cognitive:	☐	☐	__________________
Speech and language:	☐	☐	__________________
Behavior:	☐	☐	___________________
Mood:	☐	☐	___________________
Sleep:	☐	☐	___________________
Feeding:	☐	☐	___________________

Seizures: ☐ Yes ☐ No ☐ Unclear
Age of onset:
Type: ____
Controlled? ☐ Yes ☐ No
Anticonvulsants: ____

Visual Function: ☐ Normal ☐ Abnormal Age at onset of visual loss: __________
Acuity OD___________________
OS___________________
Pigmentary retinopathy: ☐ Yes ☐ No *Notes:* __________________________________
ERG: ☐ Normal ☐ Abnormal ☐ Not tested *Notes:*___________________________
VER: ☐ Normal ☐ Abnormal ☐ Not tested *Notes:* ___________________________
Visual loss characteristics: ____
____

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Neurologic Exam

Microcephaly: ☐Yes ☐No
Mental Status, describe: ____
Motor abnormality onset:______________
Motor characteristics: ____
____
Cranial nerve abnormalities: ☐Yes ☐No
Tone: ☐ Normal ☐Abnormal ________________________________
Movements: ☐Normal ☐ Abnormal ________________________________
Cerebellar ataxia: ☐Yes ☐No
Peripheral Neuropathy: ☐Yes ☐ No

Other clinical features:____

Biopsy Finding: ☐Normal ☐ Abnormal	**Cranial imaging: ☐ Normal ☐ Abnormal**
Date of test: / / *Tissue*: *EM*:	(select one) **CT scan // MRI** Date of test: / / *Findings*:

Enzyme Testing Performed:

PPT1(CLN1): ☐Normal
☐ Abnormal

TPP1(CLN2): ☐Normal
☐ Abnormal

Molecular Testing:

	Normal	Abnormal	Not Tested	
CLN1	☐	☐	☐	*N*:__________
CLN2	☐	☐	☐	*N*:__________
CLN3	☐	☐	☐	*N*:__________
CLN5	☐	☐	☐	*N*:__________
CLN6	☐	☐	☐	*N*:__________
CLN7	☐	☐	☐	*N*:__________
CLN8	☐	☐	☐	*N*:__________
CLN10	☐	☐	☐	*N*:__________

Other Medical Issues/Lab findings:

Family History: DNAshare > DNA Lab Files > Neuronal Ceroid Lipofuscinoses.doc

Recontact Info for Provider: (Name/Phone/Fax/Location/Address)

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