



Instructions

Please read carefully

- Forms must be completed by physicians, not by patients.
- **SAMPLES WILL BE ACCEPTED ONLY AFTER OUR REVIEW OF THIS INFORMATION.**
- **DO NOT SEND SAMPLES WITHOUT OUR WRITTEN REQUEST.**
- All sections must be completed. Otherwise, samples will not be accepted nor requested.
- Forms must be signed by patient/legal guardian and physician and **sent via regular mail or electronically to the address below.**
- Any information or materials submitted is voluntary and will not be returned (do not send originals).

Referring physician's name:

Specialty:

Place of active medical licensure:

Address:

E-mail:

Telephone:

Signature: _____

Patient name:

Date of birth:

Current age:

Address:

E-mail (patient, family or guardian):

Telephone (patient, family or guardian):

Signature: _____ **Name:** _____ **Relationship:** _____ **Date:** _____

Current or suspected syndromic diagnosis:

Age of onset:

How was this diagnosis established or suspected?:

What additional information is available to referring physician? (please list and date consultant reports, images, etc.):

Clinical summary (maximum 4 lines; do not send reports or images until requested):

Specific question asked of us: EVALUATION OF CLINICAL AND ANALYTIC DATA, FOLLOWED –IF APPROPRIATE AND REQUESTED BY US- BY ERYTHROCYTE GLUCOSE TRANSPORT ANALYSIS FOR THE DIAGNOSIS OF GLUCOSE TRANSPORTER TYPE 1 DEFICIENCY, AND FOLLOWED BY MOLECULAR GENETIC ANALYSIS, IF APPLICABLE.

Submission represents acceptance of these conditions:

1. The individuals, sponsoring and funding institutions, and affiliations of the Laboratories are not liable in any way from the use of any information, either requested or provided.
2. No information can be used for patient management, counseling or medical decisions unless reported in writing by us.
3. The staff of the laboratories will not become treating or consulting physicians unless explicitly stated in writing by us and their opinion will never supersede the treating or referring physicians' opinion.
4. All communications will be addressed to the referring physician.



Please complete **ALL** of the following items. See below for grading system.

	<u>YES</u>	<u>NO</u>	<u>Mild</u>	<u>Moderate</u>	<u>Severe</u>
Episodes/ Seizures	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Hypotonia/Spasticity	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Microcephaly	<input type="checkbox"/>	<input type="checkbox"/>	Head circumference:		cm; at age:
Ataxia/ Incoordination	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Articulation Difficulties	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Language Deficit	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Eye movement abnormality Specify type:	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Movement disorder Specify type: chorea, dystonia, athetosis, etc:	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Additional Comments:

DEVELOPMENTAL PARAMETERS:

Seizure Onset:

Head Growth:

Birth Weight:

Age when first walking:

Age when first speaking:

Duration of pregnancy:

Complications during pregnancy:

Apgar Scores:

Behavior:



ANALYTIC PARAMETERS:

LP Date CSF Glucose CSF protein CSF Lactate Blood Glucose Blood Lactate

OTHER NEGATIVE (NORMAL) ANALYTIC STUDIES:

NEUROIMAGING:

(Dates, modalities and results)

TREATMENT:

Date started ketogenic diet:

Date ended ketogenic diet:

Current diet:

Date and type of anticonvulsant start:

Date and type of anticonvulsant end:

Additional Comments:

CRITERIA FOR GRADING PATIENT SYMPTOMS LIST

<u>Episodes/Seizures</u>	if YES , grade:	MILD → 1x / month MODERATE → weekly or monthly SEVERE → daily or several times / week
<u>Spasticity</u>	if YES , grade:	MILD → 3+ for tendon reflexes. No ankle clonus. Mild Increase in tone. MODERATE → 4+ for tendon reflexes. Ankle clonus. Babinski signs. Increased tone. SEVERE → Spasticity. Difficulty walking/unable. 4+ tendon reflexes. Ankle clonus. Babinski signs.
<u>Ataxia/ Incoordination</u>	if YES , grade:	MILD → Incoordination. No functional impairment. MODERATE → Able to walk. SEVERE → Unable to walk.
<u>Language Deficit</u>	if YES , grade:	MILD → Speaks in sentences. Decreased vocabulary. MODERATE → Speaks in simple phrases. SEVERE → Speaks in simple words, or no language.