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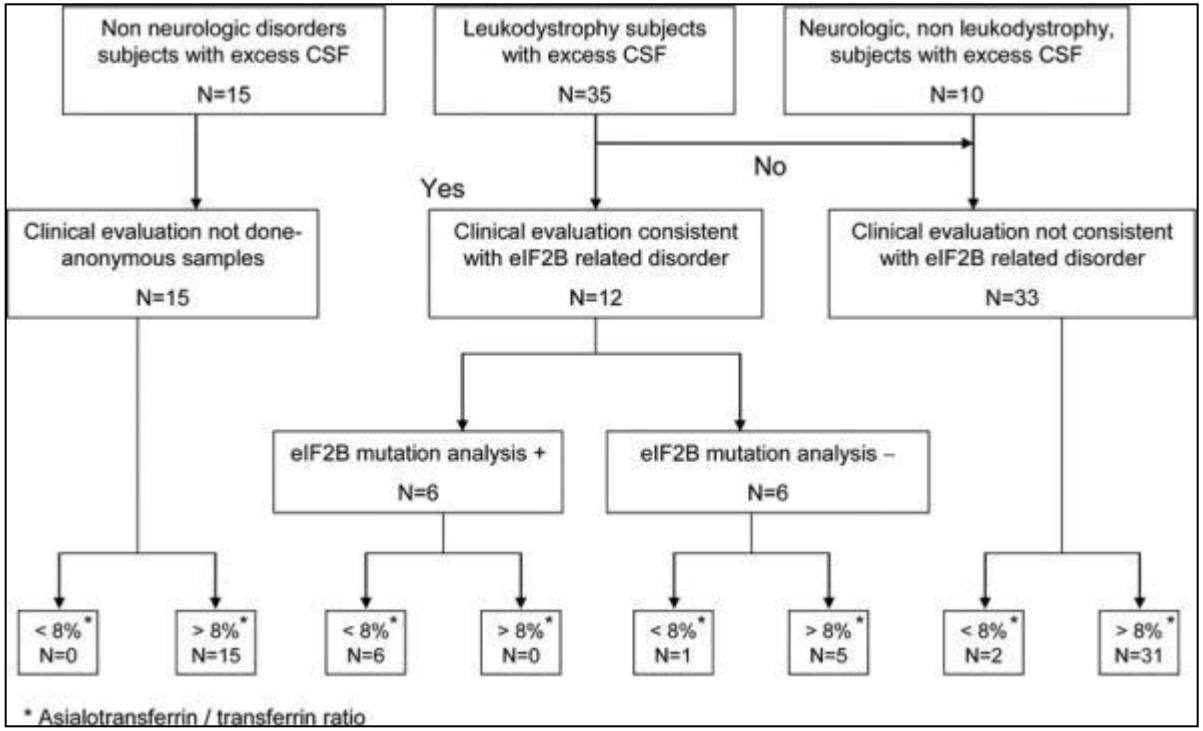
# Thank You

This PowerPoint document contains the images that you requested.

# Figure 1




**Sensitivity and specificity of decreased CSF asialotransferrin for eIF2B-related disorder.**
Vanderver, A; Hathout, Y; Maletkovic, J; Gordon, E; Mintz, M; Timmons, M; Hoffman, E; Horzinski, L; Niel, F; Fogli, A; Boespflug-Tanguy, O; Schiffmann, R

Neurology. 70(23):2226-2232, June 3, 2008.
DOI: 10.1212/01.wnl.0000313857.54398.0e

Figure 1 Flow diagram of CSF asialotransferrin/transferrin ratio diagnostic accuracy study.

# Figure 2

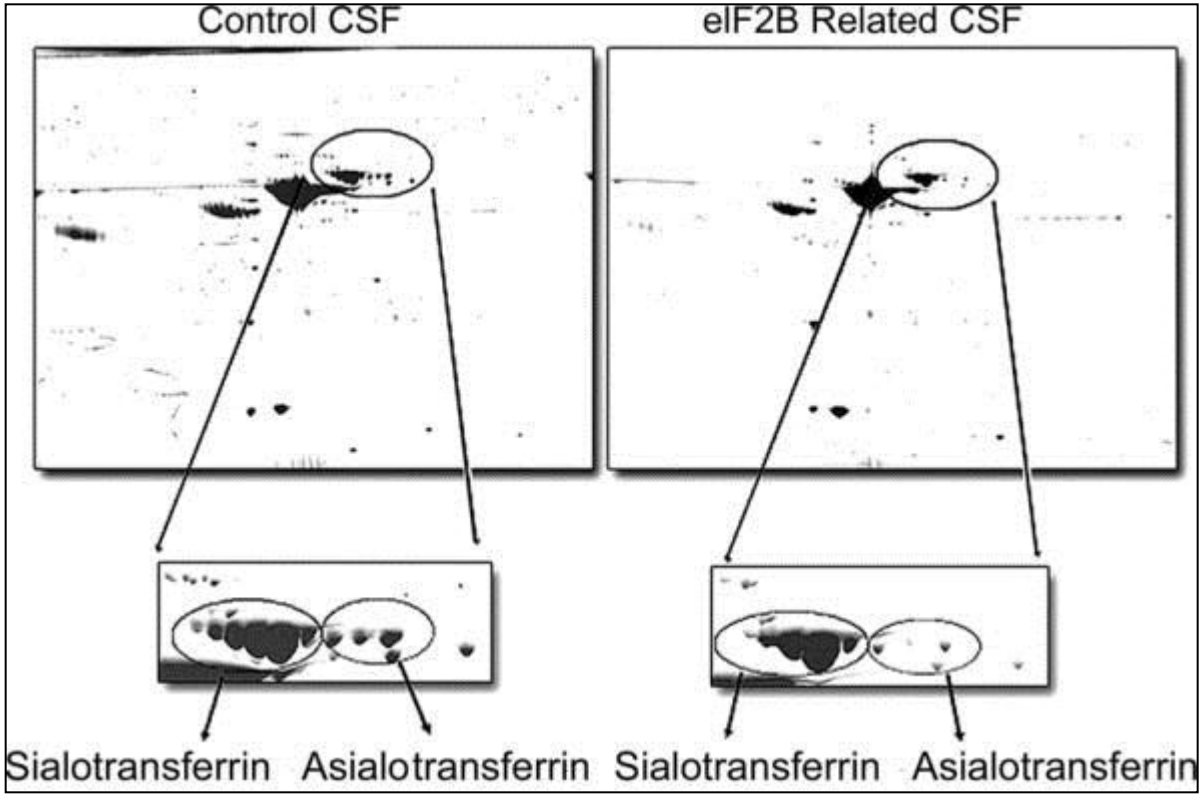



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Figure 2 Scanned image of 2DG electrophoresis of control CSF (left) and EIF2B-mutated CSF (right)Box corresponds to transferrin isoforms in each sample type. Note the decreased asialotransferrin isoforms relative to the total transferrin isoforms in EIF2B-mutated sample relative to the control sample. This patient (DMN 99.31) has mutations in EIF2B5 (Y343C/I385V).

# Table 1

Table 1 Percent of asialotransferrin to total transferrin in studied samples

| | Sample type | | | | |
|---|---|---|---|---|---|
| | *EIF2B* mutated | eIF2B-like, no mutation | Other leukodystrophy | Other neurologic | Control non-neurologic |
| Range | 0.93-7.77 | 7.98-28.92 | 5.57-48.97 | 8.63-26.98 | 10.72-30.32 |
| Mean | 4.51 | 16.46 | 15.82 | 18.87 | 18.98 |
| SD | 2.18 | 8.14 | 9.01 | 8.30 | 6.66 |

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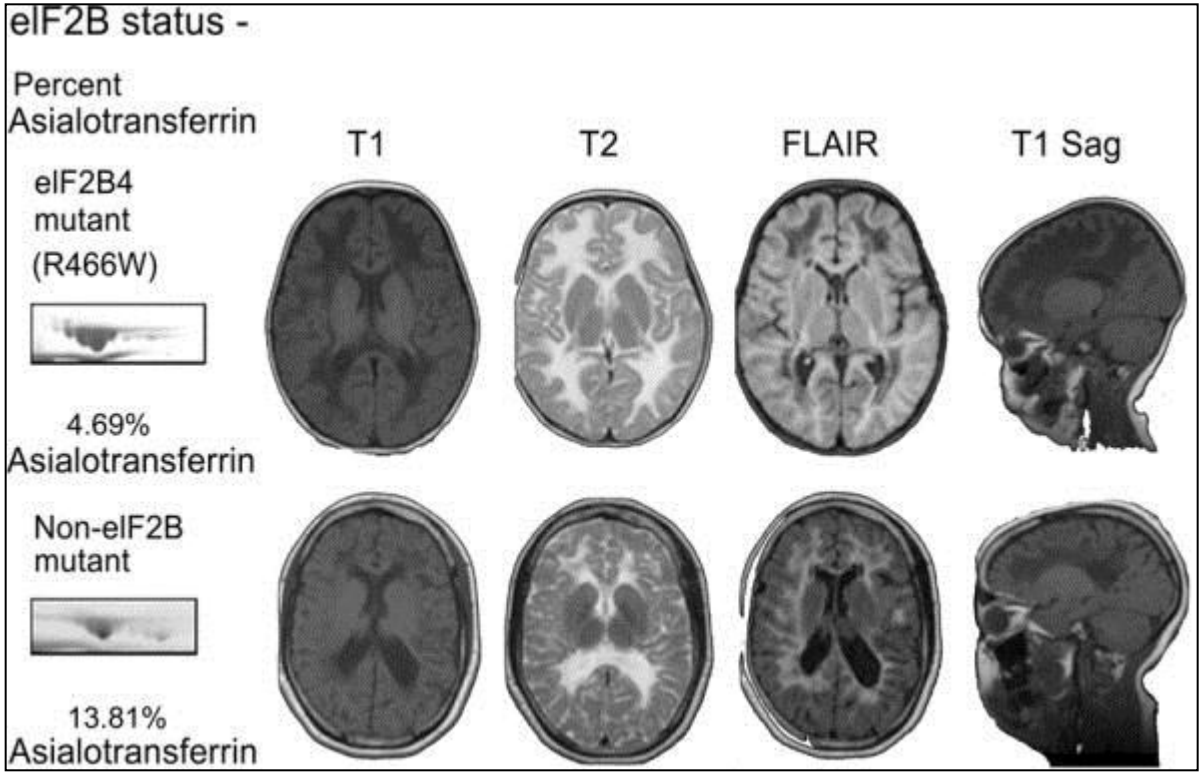


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Figure 3 Patients with EIF2B4 (R466W homozygous) mutation and with eIF2B-like clinical and radiologic presentation, but without eIF2B mutationsTop: Patient with EIF2B4 (R466W homozygous) mutation. MRI reveals T2 signal intensity similar to CSF, fluid-attenuated inversion recovery (FLAIR) images consistent with rarefaction of involved white matter, and striae on T1 sagittal images. Asialotransferrin to transferrin ratio is less than 8%. Bottom: Patient with eIF2B-like clinical and radiologic presentation, but without eIF2B mutations. MRI reveals T2 signal intensity similar to CSF, FLAIR images consistent with rarefaction of involved white matter, and striae on T1 sagittal images. Asialotransferrin to transferrin ratio is more than 8%.

# Table 2

Table 2 Clinical characteristics and mutation analysis relative to percentage of asialotransferrin in CSF in eIF2B mutated patients and eIF2B-like patients

| Patient | % Asialotransferrin | Nucleotide changes | Amino acid change | Gene | Age at disease onset | Age ay death | Symptoms | GEF activity |
|---|---|---|---|---|---|---|---|---|
| DMN 03.01 | 1.46 | 1069C>T/1069C>T | R357W/R357W | *EIF2B4* | 2 y | NA | Motor decline with febrile illness, epilepsy | NA |
| DMN 91.46 | 2.11 | 338G>A/1884G>A | R113H/W628X | *EIF2B5* | 18 mo | 6 y | Progressive ataxia after febrile illness | NA |
| DMN 96.115 | 3.71 | 512C>T/607 -612delInsTG | S171F/M203fs | *EIF2B2* | 10 y | NA | Scoliosis, gait disorder | NA |
| G648-2* | 4.11 | 638A>G/638A>G | E213G/E213G | *EIF2B2* | 7 y | NA | Progressive ataxia, behavior disturbance | 64 ± 4 |
| DMN 99.31 | 4.36 | 1028A>G/1153A>G | Y343C/I385V | *EIF2B5* | 13 mo | 5 y | Neurologic deterioration after febrile illnesses | NA |
| DMN 01.45* | 4.52 | 338G>A/splicing site exon 10 | R113H/splicing site exon 10 | *EIF2B5* | 2 y | NA | Progressive ataxia after fall | 70 ± 1 |
| DMN 91.55 | 4.65 | 338G>A/805C>T | R113H/R269X | *EIF2B5* | 18 mo | 5 y | Progressive ataxia, acute coma resulting in death | NA |
| DMN 05.06 | 4.69 | 1399C>T/1399C>T | R466W/R466W | *EIF2B4* | 15 mo | NA | Neurologic deterioration after febrile illness | NA |
| DMN 01.11* | 5.50 | 338G>A/338G>A | R113H/R113H | *EIF2B5* | 4 y | NA | Hemiparesis after fall | 77.5 ± 2.5 |
| DMN 02.02 | 7.77 | 338G>A/806G>T | R113H/R269L | *EIF2B5* | 2 y | NA | Progressive ataxia w/o provoking event | NA |
| DMNL 99.001 | 4.8 | 47C>A/338G>A | A16D/R113H | *EIF2B5* | 3 y | NA | Progressive ataxia after mild head trauma | NA |
| NA | 13.81 | None | None | None | 5 y | NA | Progressive ataxia w/o provoking event | NA |
| NA | 20.78 | None | None | None | 3 y | NA | Progressive ataxia w/o provoking event | NA |
| NA | 14.69 | None | None | None | 12 y | NA | Progressive ataxia w/o provoking event | NA |
| NA | 8.64 | None | None | None | 4 y | NA | Progressive ataxia w/o provoking event | 103.8 ± 2.8 |
| NA | 8.60 | None | None | None | 37 y | NA | Progressive ataxia w/o provoking event | 99 ± 3 |
| NA | 22.42 | None | None | None | 3 y | NA | Progressive ataxia w/o provoking event | 111.3 ± 3.7 |

*Patients previously included in Fogli et al., 2004.[27]
GEF = guanine nucleotide exchange activity, measured in lymphoblasts from affected patients; NA = not available or still alive; fs = frameshift.

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