

**48th ANNUAL DIAGNOSTIC SLIDE SESSION 2007.**

**CASE 2007 -3**

Submitted by:

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**Clinical History:** 2 week old male neonate born at 37 weeks gestational age with intractable seizures. Clinical work-up including metabolic studies and muscle biopsy all normal. History of one female and one male sibling with essentially identical clinical courses with neonatal deaths. There is one living, healthy female sibling and one living, healthy male sibling. The patient, mother and female deceased sibling all carry the same balanced  $t(8;15)(p21.1;q26.1)$ . The father and deceased male sibling have normal karyotypes. Karyotypes of the live siblings are unknown. Parents are consanguineous.

**Necropsy findings:**

3486 g male neonate

External examination: well developed neonate with large broad-based nose

Brain weight: 250 g

**Material submitted:**

- 1.) MRI at bottom of protocol
- 2.) H and E glass slide of either frontal or parietal or temporal lobe

**Points for discussion:**

- 1.) Diagnosis

