

**48th ANNUAL DIAGNOSTIC SLIDE SESSION, 2007  
DIAGNOSES AND REFERENCES**

**MODERATOR: E. Tessa Hedley-Whyte, M.D.**

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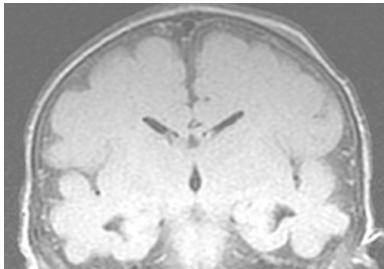
**Case 2007-3**

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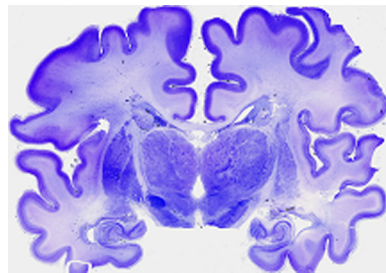
**Diagnosis: Microcephaly with simplified gyral pattern**

**Comment:** In these cases, the gyral pattern is not well formed, but there is no lissencephaly, and cortical lamination is normal. The three affected siblings in this case all had a large, flat nose, a facial feature not seen in the unaffected siblings. This appears to be an autosomal recessive disorder, with many cases occurring in families with consanguinity. Chromosomal translocations have not been described in other cases of this disorder.

From the presenter (Dr. Oviedo): The most helpful information leading to the diagnosis is the pattern of gyri seen on the MRI and the gross specimen. This pattern is clearly abnormal when compared to an age-matched control, with simplified gyri, best seen when the temporal lobes are compared. In addition, all sulci are shallow in this case. A normal control brain also has much more white matter than 2007-3. Also, it should be clearly stated that this diagnosis was made after a massive metabolic work-up that was completely negative. The gene for this entity has not yet been found.



MRI of this case: microcephaly with simplified gyral pattern.



Normal control at 37 weeks, provided by Dr. Fallet-Bianco

**References:**

Peiffer A, Singh N, Leppert M, Dobyns WB, Carey JC. Microcephaly with simplified gyral pattern in six related children. *Am J Med Gen.* 1999; 84:137-144.

Sztrihai L, Dawodu A, Gururaj A, Johansen JG. Microcephaly associated with abnormal gyral pattern. *Neuropediatrics* 2004; 35:346-352.