Schizencephaly associated with a severe prothrombotic syndrome caused by antithrombin III deficiency

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ABSTRACT

Introduction

A 12 year old boy presented with non syndromic mental retardation. Brain MRI showed schizencephaly with a deep right parietooccipital cleft extending from cortical surface to the occipital horn of cerebral ventricle. Moreover his family was known for antithrombin III deficiency linked to the homozygous c.391C>T (p.Leu131Phe) mutation in SERPINC1. Coagulation studies revealed in this young boy severe antithrombin III deficiency and molecular analysis confirmed the mutation.

Discussion

Schizencephaly can be considered as cerebral malformation of neuronal migration, caused by mutations in several transcription factors, but most cases occur sporadically and are believed to be associated with a vascular disruptive mechanism. In the proband, analysis of the SHH, SIX3 and EMX2 genes showed no mutation. Porencephaly and schizencephaly have also been attributed to mutations of COL4A1, linking, as in our case, genetic vascular pathology with schizencephaly. Otherwise mutations of methyltetrahydrofolate reductase and factor V Leiden genes seems also responsible of cases of schizencephaly. We suggest that a similar
encephaloclastic mechanism took place in our patient and hypothesize the occurrence of an early antenatal cerebral vascular injury.

**Conclusion**

Extensive coagulation studies should be performed in patients with schizencephaly before molecular analysis.

**REFERENCE**


