OP05

TCF 12 related coronal synostosis

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Introduction: The identification of *TCF12* gene mutations in uni and bi-coronal synostosis has allowed a better nosologic classification of children presenting with plagio and brachycephalies.
We reviewed the various phenotypes associated to *TCF12* mutations based on our experience at Necker Enfants-Malades, Paris.

Methods: TCF12 mutations were searched in 80 patients with coronal synostosis that were negative for FGFR3 or TWIST1 mutations.

Results: TCF12 mutations were found in 20% of the patients and most of the mutations were novel. The clinical picture of these patients was extremely variable and resembling to that of Saethre Chotzen syndrome. It could include beside the synostosis: ptosis, syndactyly, prominent ear crus, single palmar crease. In some cases these associated features were absent and the children presented only with the coronal synostosis. The clinical intrafamilial variability was important, both uni-coronal or bicoronal of cases could be found or also, in some instances, an absence of craniosynostosis could be observed.

Conclusions: This new *TCF12*-associated syndrome should be considered in children with a plagiocephaly or a brachycephaly. *TCF12* molecular testing should be realised when the molecular analysis of FGFR3 and TWIST gene is not contributory.