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UNUSUAL PRESENTATION OF COMBINED SAGGITAL-METOPIC SYNOSTOSIS MAY REPRESENT A NOVEL AUTOSOMAL DOMINANT CRANIOSYNOSTOSIS SYNDROME

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Background: Craniosynostosis, caused by early fusion of one or more cranial sutures, can include premature fusion of the saggital (scaphocephaly) or metopic suture (trigonocephaly). Though often occuring as isolated findings, their co-existence in a craniosynostosis syndrome is infrequent and mostly sporadic.

Case description: The male proband, born at term after an uneventful pregnancy, presented at birth with premature fusion of the saggital and metopic suture. Antropometric measurements were normal. Besides the scaphocephaly, a prominent boney mass on the forehead, several centimeters in height and width, was noted, next to proptosis. Imaging suggested the frontal mass to orgininate partially from the fused metopic suture, in addition to a superimposed exostosis. Radiographs did not reveal other exostoses, though bilateral agenesis of the middle phalanges in the feet was noted. Family history revealed the father, his sister and half-sister, to have an isolated scaphocephaly with variable cutaneous syndactyly. The paternal grandfather did not have craniofacial anomalies nor syndactyly, though the great grandfather was said to have isolated syndactyly of both hands.

Conclusion: This four-generation family shows various expression of a craniosynostosis phenotype with scaphocephaly and a particularly severe and unusual form of trigonocephaly. Several known craniosynostosis genes (FGFR2, FGFR3, TWIST) have already been excluded. A saggital-metopic synostosis together with agenesis of the middle phalanges has to our knowledge not been reported before. Considering the family history, this is suggestive for a novel, variable autosomal dominant craniosynostosis syndrome with possibility of gonadal mosaicism or non-penetrance.