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A Case of Craniosynostosis

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A 32-year-old pregnant woman, G2P1, spontaneously delivered a healthy male infant. At 13 weeks and 1 day of gestation, NT was 1.2 mm. At 23+4 weeks, ultrasound examination showed that BPD, HC, AC, and FL were consistent with gestational age. In the transverse plane at the level of the thalami, the hypoechoic gap between the frontal and parietal bones was absent, the hyperechoic bony ring appeared morphologically abnormal, midfacial hypoplasia was observed, and the frontal bone was prominent without depression of the nasion. The frontonasal angle was 92.45° . No syndactyly, ectrodactyly, brachydactyly, or thumb abnormalities were noted in the fingers and toes, and no proptosis was observed. NIPT at 13 weeks indicated a high risk for Muenke syndrome. Amniocentesis at 17 weeks revealed a FGFR3 gene mutation, p.Pro250Arg.

Muenke syndrome is inherited in an autosomal dominant manner with marked phenotypic variability within families. Postnatal manifestations commonly include strabismus, hearing loss, developmental delay, intellectual disability, behavioral problems, intracranial abnormalities, epilepsy, ocular abnormalities, brachydactyly, carpal and/or tarsal fusion, broad thumbs and great toes, camptodactyly, and radiographic findings such as short and broad middle phalanges and/or cone-shaped epiphyses. After genetic counseling, the woman decided to terminate the pregnancy.

