

Birth Defects, Dysmorphology, Skeletal Dysplasia, Craniofacial Anomalies

AB037. The first case of Beare-Stevenson cutis gyrata syndrome with an *FGFR2* gene mutation (Tyr375Cys) in Thailand

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Background: Beare-Stevenson cutis gyrata syndrome (BSS) is an extremely rare craniosynostosis syndrome with autosomal dominant inheritance.

Methods: We reported the first Thai case of BSS (the 27th case reported worldwide) with a heterozygous Tyr375Cys mutation in fibroblast growth factor receptor 2 (*FGFR2*) gene.

Results: A full-term Thai male infant was presented with bilateral ventriculomegaly and Chiari malformation type II detected by prenatal ultrasonography. The classic clinical features of BSS including clover-leaf skull shape, ocular hypertelorism, proptosis, cutis gyrata at pre-auricular area,

natal teeth, prominent umbilical stump, glandular type hypospadias with bifid scrotum, anteriorly placed anus, and deep skin furrowed of both palms and soles were found on physical examination at birth. The diagnosis of BSS was made based on typical clinical features and confirmed by detection of heterozygous Tyr375Cys mutation in *FGFR2* gene. He had severe birth asphyxia and developed respiratory failure shortly after birth. According to severe multiple organs involvement and poor clinical outcome, palliative care was offered with parental agreement. The baby died at the second day of life.

Conclusions: BSS should be considered in an individual with craniosynostosis accompanied with cutis gyrata. Confirming the diagnosis by *FGFR2* mutation analysis is highly recommended for genetic counseling and determining the prognosis.

Keywords: Beare-Stevenson cutis gyrata syndrome; craniosynostosis; fibroblast growth factor receptor 2 (*FGFR2*)

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