Nannaphat Atsawaphidsawat et al., J Clin Exp Pathol 2017, 7:5 (Suppl)
DOI: 10.4172/2161-0681-C1-041

Beare-Stevenson cutis gyrata syndrome with full body autopsy: A case report with vascular abnormalities

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Introduction: Beare-Stevenson cutis gyrata Syndrome (BSS) is a seriously extremely rare genetic disorder characterized by skin abnormalities and the premature fusion of certain bones of the skull (craniosynostosis). Recently evidence was presented that BSS is caused by a mutation of the *FGFR2* gene. Here, we demonstrate the first Thai case of BSS (the 27th case in English publication) with full body autopsy.

Case Report: A full-term Thai male infant, without any evidences of congenital anomalies in his family, was found having bilateral ventriculomegaly during intrauterine period. Soon after birth, the baby developed cyanosis and immediate intubation was given. His condition became worsen, until his death in second day of life. Consent for full body autopsy was given by the family members. External examination revealed classical features of BSS including clover-leaf skull shape, cutis gyrata, prominent umbilical stump, ambiguous genitalia, but acanthosis nigricans was not detected. Internal examination revealed craniosynostosis, Arnold-Chiari malformation type II, hydrocephalus and agenesis of corpus callosum. Interestingly, microscopic study of skin at the nape of neck showed glomuvenous malformation and glomangiomyoma-like lesions that was the first perivascular tumor described in BSS. Molecular analysis of *FGFR2* gene confirmed a heterozyguous p.Tyr375Cys, which was identical to that detected in previous 14 cases.

Conclusion: BSS should be considered in patients presented with craniosynostosis and cutis gyrata. Associated vascular malformations in BSS were first described in this case. The further study of additional patients will provide more information about the clinical phenotypes.

Biography

Nannaphat Atsawaphidsawat has completed her MD from Khon Kaen University in 2016. She has then continued her specialty in Anatomical Pathology, Faculty of Medicine, Khon Kaen University, Thailand.

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