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

Nannaphat Atsawaphidsawat,
Khon Kaen University, Thailand

ABSTRACT

Presentation: Beare-Stevenson cutis spin Syndrome (BSS) is a genuinely very uncommon hereditary issue portrayed by skin variations from the norm and the untimely combination of specific bones of the skull (craniosynostosis). As of late proof was introduced that BSS is brought about by a change of the FGFR2 quality. Here, we exhibit the main Thai instance of BSS (the 27th case in English distribution) with full body post-mortem examination.

Case Report: A full-term Thai male new-born child, with no confirmations of inherent abnormalities in his family, was found having two-sided ventriculomegaly during intrauterine period. Not long after birth, the child created cyanosis and quick intubation was given. His condition became decline, until his passing in second day of life. Assent for full body examination was given by the relatives. Outer assessment uncovered old style highlights of BSS including clover-leaf skull shape, cutis spin, unmistakable umbilical stump, equivocal genitalia, yet acanthosis nigricans was not distinguished. Interior assessment uncovered craniosynostosis, Arnold-Chiari distortion type II, hydrocephalus and agenesis of corpus callosum. Strikingly, minuscule investigation of skin at the scruff of neck demonstrated glomuvenous distortion and glomangiomyoma-like injuries that was the first perivascular tumor depicted in BSS. Atomic investigation of FGFR2 quality affirmed a heterozygous p. Tyr375Cys, which was indistinguishable from that distinguished in past 14 cases.

End: BSS ought to be considered in patients gave craniosynostosis and cutis gyrata. Related vascular distortions in BSS were first depicted for this situation. The further investigation of extra patients will give more data about the clinical phenotypes.

nnpats@gmail.com

Note: This work is partly presented at 14th Asia Pacific Pathology Congress, NOVEMBER 13-14, 2017 | OSAKA, JAPAN.