



Response to Letter to the Editor: Giant Cardiac Rhabdomyoma with Mixed Atrial Tachycardia and Nonsustained Ventricular Tachycardia in a Newborn with Tuberous Sclerosis

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Tuberous sclerosis is rare multisystemic genetic disease caused by mutations of TSC1 or TSC2 and characterized by tumor-like manifestations that involve the brain, heart, kidneys, lungs, eyes and skin. Cardiac rhabdomyomas are the most common cardiac feature of the tuberous sclerosis in the fetus and neonate. It is reported that 60%–80% of children affected by tuberous sclerosis have cardiac rhabdomyomas [1].

In the case, we demonstrated the multiple systemic involvement related to tuberous sclerosis [2]. Brain ultrasound showed multiple cortical and subcortical tubers on both fronto-parieto-occipital area. But, brain MRI was not performed because unstable hemodynamic state with continued ventilator therapy during admission. In addition to cardiac rhabdomyomas, the patient also had renal malformations. Kidney ultrasound demonstrated that several tiny renal cyst in both kidneys after postnatal examination. The spongiform shaped cystic lesion in lower portion of right kidney and clustered cystic lesion in upper portion of left kidney, which was probable finding due to tuberous sclerosis involvement.

Maternal, perinatal and familial risk factors with tuberous sclerosis are associated with the postnatal prognosis of cardiac rhabdomyomas [3].

In the case, the patient was born by in vitro fertilization at a gestational age of 36 weeks. The patient was first baby and there was no siblings. Regarding the maternal history, the mother's age was 32 at the time of delivery. The mother had negative serological tests for HIV, syphilis, and hepatitis B. And no other medical history had been documented. There was no complication during pregnancy. Although the preterm labor accompanied at 32 week of gestation, it was recovered soon. There was no relevant family history of dermatological lesion and seizure.

As the author pointed out, the tuberous sclerosis is inherited in an autosomal dominant manner with high penetrance. Therefore parental genetic examination is also required for establishing whether the disease is inherited or occur sporadically. In the study, genetic testing of the parents showed no abnormalities.

Unfortunately, no autopsy was performed after the patient died. Parents disagreed because of the negative cultural perception of autopsy. As the author noted, the histological finding of

cardiac tumors may have identified the any pathological findings related to tachyarrhythmias and the cause of the fatal outcome of the patient.

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Not applicable.

Conflict of Interest

No potential conflict of interest relevant to this article was reported.

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Author Contribution

The article is prepared by a single author.

Ethics Approval and Consent to Participate

Not applicable.

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