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A Case Report on a Neonate diagnosed with Complete Penile Agenesis or Aphallia

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Abstract

Background: Complete penile agenesis (Aphallia) is a very rare congenital anomaly, with an incidence of about one in 30 million births. It is believed to result from the absence or a failure in the development of the genital tubercle. Diagnosis of this rare anomaly is made by clinical examination, but treatment options and parental counselling for gender assignment is difficult and challenging for the treating doctor.

Objectives: To present a case of a neonate diagnosed with Aphallia or Complete Penile Agenesis (CPA)

Case: A male neonate born via normal spontaneous delivery to a mother who had an unremarkable prenatal history. At bith, he presented with a complete absence of a phallus with descended testes bilaterally. Ultrasound of the the abdomen was done with unremarkable results. Patient was refered to a team of specialists and the following work-up were suggested: baseline testosterone levels, HCG stimulation test and karyotyping. Patient was discharged with plans for a multidisciplinary team approach, and appropriate parental counselling to help in reducing psychosocial problems.

Conclusion: Infants diagnosed with Aphallia requires an appropriate parenteral counselling can assist the family in reducing psychosocial problems. Treatment of CPA presents many challenges involving a multidisciplinary approach. The team should include a urologist, a pediatrician, an endocrinologist, a geneticist, and a mental health expert. Management requires emphasis on two therapeutic methods: feminizing genitoplasty and phalloplasty.

Biography:

Jabez Tuñacao has completed his medical education at the age of 24 years at Cebu Doctors University and is currently training as a pediatric resident at Perpetual Succour Hospital in Cebu City.

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