

VICI'S SYNDROME ASSOCIATED TO CLEFT PALATE: CLINICAL REPORT ON A BRAZILIAN BOY

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OBJECTIVE: to report on a boy with Vici's syndrome associated to cleft palate. **CLINICAL REPORT:** JCH, male, the 4th child born to normal and non-consanguineous parents, pregnancy and delivery were unremarkable. Neonatal period was complicated by jaundice that was responsive to phototherapy. He also presented feeding problems and cyanosis when he was crying. Genetic evaluation performed at 20 days of life showed albinism, large head, and cleft palate. Routine laboratory tests were unremarkable, G-banded chromosomes in peripheral lymphocytes showed a normal 46, XY constitution. Brain MRI showed agenesis of the corpus callosum and hydrocephalus. Hearing evaluation showed sensorineural loss. At age 18 months he was submitted for cleft palate surgery repair. Re-evaluation at age 3 years showed severe cognitive deficit, aggressive behavior and he was starting to walk with support. The presence of callosal agenesis, severe psychomotor retardation, oculocutaneous albinism, and hearing impairment was consistent with the diagnosis of Vici's syndrome, a very rare autosomal recessive disorder whose gene is hitherto undefined. **CONCLUSIONS:** here we report on a patient presenting the main signs of the Vici's syndrome. This condition has been seldom reported and to our knowledge cleft palate was only reported once. Multisystem previously reported such as cardiomyopathy, neuromuscular and immunological disturbances corroborate for the heterogeneity existing within the spectrum of this rare condition. Despite to be rare, more cases are needed to establish the minimal criteria to the understanding of the genetics and molecular basis of this condition.