

A case with 47,XXY,del(11)(q23) karyotype-coexistence of Jacobsen and Klinefelter syndromes

Abstract

A case with 47,XXY, del(11)(q23) karyotype-coexistence of Jacobsen and Klinefelter syndromes: A two-year-old dysmorphic male child was found to have 47,XXY,del(11)(q23) karyotype. Domination of the clinical features of Jacobsen syndrome was observed: mild mental retardation, trigonocephaly, ptosis, downward slanting palpebral fissures, low set ears, carp-shape mouth and micrognathia. Transient thrombocytopenia and leukopenia were also present. Over the following five years gynecomastia and eunuchoid body proportions became evident as clinical features of Klinefelter syndrome. This is the first description of the coexistence of both syndromes.

Matheisel A, Babinska M, Wierzba J, Wozniak A, Nedoszytko B, Balcerska A, Limon J. A case with 47,XXY,del(11)(q23) karyotype-coexistence of Jacobsen and Klinefelter syndromes. *Genet Couns.* 2000;11(3):267-71. PMID: 11043435.