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Abstract

Immense recidive two-sided inguinal hernias in a tetrasomy 12p disorder.

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Abstract:

Aim: Immense recidive two-sided inguinal hernias in a tetrasomy 12p disorder..

We present an aterm male (Birth weight: 3860 gr) (AD 39 6/7 weeks PML) of a G8P7AB 1 of a 42 year old Moroccan mother(consanguinity).The prenatal ultrasound demonstrated an unilateral ventriculomegaly and polyhydramnios.After birth we found an extremely hypotonic male newborn with characteristic facial features include a high, rounded forehead; a broad nasal bridge; a short nose; widely spaced eyes; low-set ears; rounded cheeks; and a wide mouth with a thin upper lip a large tongue and a high arched palate.Feeding difficulties were present: GERD,

swallow fifficulties, constipation and micro-and microaspirations (fever and respiratory infections). The genetic diagnosis confirmed a Pallister-Killian syndrome The SNP array showed a pathogenic multiplication of the entire arm of chromosome 12p. This fits with the diagnosis of a Pallister-Killian syndrome (tetrasomy 12p) At the age of 2 months he developed huge bilateral inguina-scrotal hernias.An ultrasound showed the presence of intestinal structures visible in the canalis inguinalis and more caudally in the tunica

vaginalis, reaching as far as intrascrotal on the left and as far as the entrance of the scrotum on

the right side. At the age of 4 months an hernioraphy bilateral was done. Afterwards there was a recidive at the age of 11 months with a laparoscopic Pallister-Killian syndrome is a dysmorphic condition involving most organ systems, but also characterized by a tissue-limited mosaicism; most fibroblasts have 47 chromosomes with an extra small metacentric chromosome, whereas the karyotype of lymphocytes is normal. The extra metacentric chromosome is an isochromosome for part of the short arm of chromosome 12.

Biography:

Dr. Martine Docx, Working on Department of Chronic Paediatric Diseases and Eating Disorders Chronic disease in children especially hypertension,nephrology and eating disorders Queen Paola Children's Hospital Antwerp Belgium

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