stimulating hormone, luteinizing hormone, and total testosterone levels were 33.02 mIU/mL (normal reference range <6.7), 0.54 mIU/mL (normal reference range 0.3-6.0), and 5.19 ng/dL (normal reference range <7), respectively. Serum testosterone levels were not increased in response to 1500 units/dose HCG stimulation test for 3 days. The diagnosis of vanishing testis was confirmed and hormone replacement therapy was planned to start during puberty.

Early diagnosis and appropriate treatment will prevent the development of hypogonadism complications.

(P-46)

Warburg Micro Syndrome: A New Case from Consanguineous Parents

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Warburg micro syndrome (WARBM) is a rare autosomal recessive disorder characterized by postnatal severe intellectual disability, microcephaly, ocular findings such as congenital cataract, microcornea, microphthalmia and optic atrophy, and hypogonadism. *RAB3GAP1* mutations are mostly causative. This child was presented because of having two patients from the same family. First son of the family had WARBM and he died.

A 12-month-old boy was admitted to our clinic because of congenital cataracts, micropenis, and cryptorchidism. Parents were second cousins. Birth weight was 2800 g. At 2 months of age, he underwent bilaterally phacoemulsification. His weight was 6.8 kg (<3p), length 75.5 cm (25-50p), and head circumference was 41 cm (<3p). He had low-set and large ears, a prominent nasal root, ptosis, high-arched palate, and micrognathia. Ophthalmological examination revealed hypotelorism, microphthalmia, and microcornea. He had truncal hypotonia, increased muscle tone in both legs, poor head control, and was unable to sit without support. He had bilateral cryptorchidism, a micropenis (stretched penile length of 20 mm), and scrotal hypoplasia. USG revealed both testicles within the inguinal canals. Karyotype was 46, XY, inv(9)(p12q13). Failed to get visual message as a result of Flash VEP. Cranial MRI showed atrophy of the corpus callosum and cerebral cortex. Sequence analyses of the *RAB3GAP1* gene revealed that he was homozygous for the splice donor mutation c.748+1G>A. His deceased brother possessed the same mutation. Both parents were heterozygous for the mutation.

WARBM is rarely presented in siblings. The importance of pre-conception genetic counseling in the family who has children with same disorders is highlighted.

(P-47)

Williams Syndrome Associated with Isolated Growth Hormone Deficiency: Is It Just a Coincidence?

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Williams-Beuren syndrome (WBS) is a rare disorder caused by chromosome 7q11.23 deletion. Clinical manifestations are a happy looking dysmorphism, moderate mental retardation, growth retardation, and congenital heart defects (CHD). Short stature is found in about 50% of children with WBS, however, it is generally not severe. Here, we report a case of WBS associated with growth hormone deficiency (GHD).

The patient was the first child of healthy, non-consanguineous parents. He was born at term with weight of 2500 g (3p), length 48 cm (3p), and head circumference (HC) of 33 cm (3p). At age 3 years, he was referred for the first time because of dysmorphism and CHD. Serum calcium levels were elevated. He had severe growth retardation. His height was 73.6 cm (≤3p), weight 7300 g (≤3p), and HC was 43.5 cm (below -2 SDS). The karyotype was normal. FISH analysis showed hemizygously deleted 7q11.23. Measures yielded during endocrine evaluation were indicative of severe GHD. Treatment with hGH 1 U/day was started to which our patient responded well.

Short stature is found in about 50% of children with WBS. It is usually not severe and the postnatal overall growth is frequently along the 3p. For some WBS patients on the contrary, growth retardation is severe. To the best of our knowledge, there are only 3 patients with WBS reported to have associated GHD. The pathogenesis of GHD is unclear. A hypothalamic rather than pituitary defect is suggested. We recommend evaluation of growth hormone or at least screening by IGF-I measurements in all patients with microdeletions and unexplained short stature.