her genetic potential (mother’s height 159.5 cm, father’s height 176 cm, mid parental height 162.5 cm)

Conclusion Topical ocular administration of corticosteroid preparations, although rarely, may lead to the development of IAL. Growth suppression due to corticosteroid administration may occur in children without other symptoms of Cushing syndrome. Growth monitoring is required in all children receiving long-term topical corticosteroid therapy, and discontinuation of therapy should be gradual due to the possibility of developing of an adrenal crisis.

46,XY DISORDER OF SEX DEVELOPMENT – PARTIAL GONADAL DYSGENESIS – CASE REPORT


Introduction 46, XY partial gonadal dysgenesis (PGD) is a disorder of sex development characterised by an incomplete testicular development (dygenetic gonad) which results in incomplete virilisation of external genitalia in utero and partial involution of Müllerian ducts in individuals whose karyotype is 46,XY.

Case Report A 14-year old girl was admitted due to primary amenorrhoea. A physical examination showed: BW 66 kg (88. c.), BH 156.7 cm (22. c.), BMI 24.9 kg/m² (94. c.), axillary hair (Tanner stage 4), the lack of breast development (Tanner stage 1), clitoromegaly/micropenis (length of 3 cm) with the urethral opening at its tip, labial fusion, no palpable gonads; the rest of the physical examination was without abnormalities. She referred to herself as a female.

The results of the diagnostic evaluation: high gonadotropin levels – LH (11.6 IU/L) and FSH (58.6 IU/L), low levels of sex hormones – testosterone (0.6 nmol/L) and oestradiol (<32 nmol/L), low AMH (<0.21 nmol/L) and normal androstenedione (4.3 nmol/L) and 17-OHP (2.1 nmol/L). Karyotype was 46,XY with partial duplication Xp ((46,XY dupl(X) (p11.4p22.1)). There was no increase in plasma testosterone (811 IU/L; ref. <5) and total testosterone (82.2 nmol/L; ref. 0.8-1.3), normal concentrations of alpha-fetoprotein, DHEAS, androstenedione and 17-OHP, immeasurably low concentrations of LH and FSH. Bone age assessment by Greulich-Pyle atlas was 13.5 years. Brain MRI: expansive lesion in the lower part there was a vagina which measured 8.5 cm in length, closed at the end. Surgical exploration and gonadectomy were performed. Pathohistological evaluation of the gonads showed structures of incompletely developed testis – clusters of incompletely developed convoluted seminiferous tubules with the absence of germinative cells, single cell atrophy and sclerosis of tubules, with hyperplasia of Leydig cells and persevered tubules of rette testis, as well as all efferent ductules of the testis; there was no tumour tissue. Karyotype of the gonads matched the one from the peripheral blood – 46,XY dupl(X)(p11.4p22.21). Oestrogen substitution was initiated.

Discussion Patients with 46, XY PGD generally have their diagnosis established shortly after birth during evaluation of the ambiguous genitalia. In a smaller number of girls, as was in our patient, it is revealed during puberty due to the lack of anticipated sex development. Even though it is indisputable, genetic background is unknown in more than 30% of patients. Alongside with 46,XY karyotype, our patient had duplication of Xp(p11.4p22.1). Duplicated region contains NR0B1 gene, which has an important role in the process of sex differentiation; furthermore, changes in the number of its copies are described in some of the patients with 46,XY gonadal dysgenesis. On the subject of patients with 46,XY PGD, it is important to establish the diagnosis as soon as possible regarding the malignant potential of the dysgenetic gonads and the need of prophylactic gonadectomy.

PATTERNS OF TSH AND GONADOTROPINS IN PATIENTS WITH SEVERE, TREATED PRIMARY HYPOTHYROIDISM

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Introduction Hypothyroidism in children causes growth retardation and delayed puberty, but in some cases long-lasting and severe, untreated primary hypothyroidism can cause precocious puberty and hyperprolactinemia with or without galactorrhea – Van Wyk-Grumbach syndrome (VWGS). It is hypothesized that ‘hormonal overlap’ of TSH and gonadotropins who