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 IAI. 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.

220 46,XY DISORDER OF SEX DEVELOPMENT – PARTIAL GONADAL DYSGENESIS – CASE REPORT

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10.1136/archdischild-2021-europaediatrics.220

Introduction 46, XY partial gonadal dysgenesis (PGD) is a disorder of sex development characterised by an incomplete testicular development (dysgenetic 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 amenorrhea. A physical examination showed: BW 66 kg (88. c.), BH 156.7 cm (22.c.), BMI 24.9 kg/m² (94.c.), axillary and pubic 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 following hCG administration. Pelvic MRI showed no uterus, vagina was positioned commonly with blind ending, structures resembling dysgenetic gonads were found bilaterally at the level of internal inguinal rings. Cystovaginoscopy revealed an opening of the short urogenital sinus beneath the hyperplastic clitoris/hypoplastic penis, with the upper part opening at the urinary bladder and the normoposition of the orifice; at 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 rete 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)(p11.4p22.1). 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.

221 PRECOCIOUS PUBERTY CAUSED BY GERM CELL TUMOR OF THE CENTRAL NERVOUS SYSTEM – CASE REPORT

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10.1136/archdischild-2021-europaediatrics.221

We present a boy with peripheral precocious puberty caused by germ cell tumor of the central nervous system. Our patient presented at the age of 10.5 years with accelerated growth and progressive virilization in the preceding 6 months (he gained about 11 cm in height, his muscle mass increased significantly, accelerated genital development was noticed, as well as acne occurrence and greasy hair). There were no headaches or visual disturbances, and he did not complain of frequent urination. Physical examination: BW 52 kg (96th ct.), BH 152 cm (93rd ct.), prominent muscles, genitals stage 4 on Tanner scale. Laboratory findings: increased concentrations of βhCG (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 pineal region with radio-morphological characteristics of non-germinomatous germ cell tumor (NGGCT) – choriocarcinoma (according to laboratory findings). After chemo and radiotherapy remnants of the tumor were surgically removed (histopathology of previously irradiated tissue was noninformative). Most cases of precocious puberty are seen in girls, in whom it is usually idiopathic. In boys, however, it is often caused by an underlying disease which is why it should always be taken seriously and investigated in a timely manner.

222 VAN WYK-GRUMBACH SYNDROME – A RARE MANIFESTATION OF UNTREATED PRIMARY HYPOTHYROIDISM

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10.1136/archdischild-2021-europaediatrics.222

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