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.

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

Matea Melša*, Miram Pasini, Nevena Krnić, Marija Mikloš, Kristina Crkvenac Gornik, Ivana Sabolić, Katja Dumić Kubat. *Dom zdravlja Zagreb – Centar*

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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.

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

Duje Braovac*, Filip Jadrijević-Cvrlje, Vesna Kušec, Hrvoje Jednačak, Miroslav Dumić. *Zavod za dječju endokrinologiju i dijabetes, Klinika za pedijatriju KBC Zagreb*

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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 β hCG (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 assesment 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.

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

¹Maja Vinković*, ^{1,2}Nevena Krnić, ¹Anita Špehar Uroić, ^{1,2}Vesna Kušec, ¹Duje Braovac, ^{1,2}Katja Dumić Kubat. ¹Department of Pediatrics, University Hospital Centre Zagreb; ²School of Medicine, University of Zagreb

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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

contain an identical alpha chain leads to stimulation of gonadal FSH receptors by supraphysiological TSH concentrations.

Case Report We present a 8.5-year-old girl referred to endocrinologist due to obesity and growth retardation that have worsened in the past year. Medical history revealed breast growth without pubic and axillary hair at the age of 8 years and menarche at the age of 8.5 years. Physical examination: short stature and generalized obesity (height 110.2 cm, - 3.6 SD; weight 40.3 kg, +1.8 SD), bradycardia (58 bpm), edematous and dry skin, thin hair, hoarse voice, thelarche Tanner stage III and absence of galactorrhea, pubic and axillary hair. The thyroid gland was not palpable. Laboratory tests revealed severe primary hypothyroidism as part of autoimmune thyroiditis (fT4 5.2 nmol/L, ref. 11-19.5; TSH > 100 mIU/L, ref. 0.05-4.8; anti-TPO 250 IU/mL, ref. < 4.3). US: hypoechoic, hypotrophic thyroid without focal changes. Elevated estradiol (72 pmol/L, ref. < 32 pmol/L) and prolactin concentrations (45 ug/L, ref. < 20 ug/L) with normal gonadotropins were detected. Other significant laboratory findings were macrocytic anemia, hypercholesterolemia and elevated CK and LDH concentrations. X-ray of the wrist showed a bone age of 6 years. Pelvic US: enlarged uterus with thickened endometrium (5 mm) and bilaterally enlarged multicystic ovaries.

Conclusion A diagnosis of VWGS was established and L-thyroxine replacement therapy was introduced. It resulted in normalization of laboratory findings, cessation of menstruation and regression of breast glandular tissue. In the following year her height increased for 15 cm and weight was reduced for 10 kg.

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EXTREME GROWTH RETARDATION CAUSED BY PRIMARY HYPOTHYROIDISM

G Krajnović*, D Mitrović Dittrich, M Stanić, I Krajnović. OB „Dr.Josip Benčević“ Slavonski Brod

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A twelve-year old girl was hospitalized because of extremely short stature. She was born from normal pregnancy and delivery weight was 3000 g and height 51 cm. She was not ill. Her psychomotoric development was normal. At the age of seven she was 115 cm tall (3. ct), and now, at the age of twelve 118.5 cm (Z score -4.64). She did not visit her doctor at that period. She was extremely short, her skin was dry, she was amimic, bradycardial, she had slowed and irregular tooth growth, and she did not have any signs of pubertal development.

In laboratory test: fT4 <0.4 ng/dl, fT3-<0.1 pg/ml, TSH>100 uIU/ml, high levels of cholesterol, anemia, high levels of AST, ALT, CK, pleural and pericardiac effusion. Bone maturity matched that of a six-year old. ITT showed low production of growth hormone, and MRI of hypothalamic pituitary region was normal. We started treatment with levothyroxine, 50 ug per day, and vitamin D. After a few months her laboratory tests were normal. In the period of one year she has grown 11.5 cm, she has gained 5 kg, her bone maturity has improved and now matches that of a ten-year old, puberty started, her skin was normalized, her mood

has improved, she is smiling more often and achieves better results at school. Her family is not cooperative so we included Social Welfare. We follow her development continuously

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IMPAIRED SLEEP QUALITY IN CHILDREN WITH INFLAMMATORY BOWEL DISEASE PRESENT EVEN IN THE REMISSION PHASE AND ATTRIBUTING TO IMPAIRED HEALTH RELATED QUALITY OF LIFE

Ivana Trivić*, Zrinjka Mišak, Sanja Kolaček, Iva Hojsak. Referral Centre for Paediatric Gastroenterology and Nutrition, Children's Hospital Zagreb

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Children with inflammatory bowel disease (IBD) have significantly lower health related quality of life (HRQoL) compared to healthy controls. HRQoL presents a broad, multidimensional concept compromising one's physical health, psychological state, level of independence, social relationships, personal beliefs and relationship to the environment. Good sleep is essential in maintaining health and quality of life (QoL) and plays a role in regulation of immune and neuroendocrine system. The aim of our study was to evaluate the relationship between sleep quality and HRQoL in children with IBD in remission.

A total of 33 paediatric IBD patients in remission (20 boys) aged 15.6 ± 1.9 years were included in the study (disease type: Crohn's disease (CD), n=16, ulcerative colitis (UC), n=15, inflammatory bowel disease-unclassified (IBD-U), n=2). Sleep quality was assessed using the Pittsburgh Sleep Quality Index (PSQI) questionnaire, whilst HRQoL was assessed using IMPACT III questionnaire. Moreover, patients wore a triaxial accelerometer for five consecutive days for objective PA quantification. Anthropometric data and inflammatory markers' values such as C-reactive protein (CRP), erythrocyte sedimentation rate (ESR) and faecal calprotectin values were recorded.

Prevalence of impaired sleep quality (PSQI>5) was 36.4%, with mean PSQI score 4.64 ± 2.21 . Highest mean scores were recorded in the sleep duration (mean score 1.06 ± 0.99), sleep disturbance (mean score 1.06 ± 0.35) and daytime dysfunction (mean score 1.00 ± 0.79) components of the questionnaire.

Mean IMPACT III score was 146.36 ± 17.24 . On average, patients spent 38 minutes in moderate-to-vigorous physical activity (MVPA), and 198 minutes in light physical activity (LPA) per day. PSQI score negatively correlated with IMPACT III score (coef. -0.446, $p < 0.01$); meaning that the more significantly impaired sleep quality the more impaired QoL; and with time spent in LPA (coef. -0.482, $p < 0.01$). Interestingly, faecal calprotectin only positively correlated with sleep disturbance score (coef. 0.352, $p = 0.048$), but had no significant correlation with the total PSQI score. No correlation was found between anthropometric and other laboratory parameters, MVPA and PSQI and IMPACT III scores.

More than a third of paediatric IBD patients suffer from poor sleep quality even in the remission phase. Further studies