and an MODY diabetes. At present blood glucose and glycated hemoglobin are normal, renal function is stable.
Conclusions: HNF1β mutation is well known to nephrologists we must not forget that the gynecological examination is essential in the context of associated malformations.

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A new case of Bardet-Biedl syndrome associated with vaginal atresia and uterus hypoplasia

Leyla Akin[1]; Selim Kurtoglu[1]; Mustafa Kendirci[1]; Mustafa Kucukaydin[2]
[1]Erciyes University Faculty of Medicine, Pediatric Endocrinology, Kayseri, Turkey; [2]Erciyes University Faculty of Medicine, Pediatric Surgery, Kayseri, Turkey

Background: Bardet-Biedl syndrome is an autosomal recessive disorder characterized by retinal dystrophy, digital malformations, obesity, mental retardation, hypogonadism (described mainly in males) and renal anomalies. Genital abnormalities in females with Bardet-Biedl syndrome have been rarely reported, including hypoplasia of uterus, ovaries, and fallopian tubes, uterus duplex, vaginal atresia and septate vagina. Most of these anomalies were missed in the childhood.
Case: A 15 year-old female with Bardet-Biedl syndrome was presented to our clinic due to evaluation of primary amenorrhea. The pubertal status was stage 4 according to Tanner staging. Pelvic ultrasonography showed uterus hypoplasia and normal ovaries. Genitogram revealed vaginal atresia. Reconstructive surgery was planned.
Conclusion: This patient was reported to emphasize the possibility of association of genital anomalies in females with Bardet-Biedl syndrome. Early systematic evaluation of patients with this syndrome for genital anomalies will prevent late diagnosis and possible complications.

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Endocrine profile, BMD evaluation, estroprogestinic treatment in the follow up of girls with congenital coagulopathies

Maria Cristina Maggio[1]; Fabio Gagliano[2]; Giuseppe Salvo[3]; Eugenia Prinzi[3]; Silvano Bertelloni[4]; Giovanni Corsello[5]
[1]University of Palermo, Universitary Department "Materno-Infantile, of Andrology and Urology", Palermo, Italy; [2]ARNAS Civico, Palermo, IV Maggiore, Palermo, Italy; [3]University of Palermo, Palermo, Italy, Universitary Department "Materno-Infantile, of Andrology and Urology", Palermo, Italy; [4]Pediatric Endocrinology, Division of Paediatrics, Department of Reproductive Medicine and Paediatrics, University of Pisa, Pisa, Italy; [5]University of Palermo, Palermo, Italy, Universitary Department "Materno-Infantile, of Andrology and Urology, Palermo, Italy

Background: Menarche is a crucial event in the life of adolescents affected by coagulopathies, with a risk of a severe blood loss if not adequately and promptly treated since menarche.
Objective and hypotheses: A multidisciplinary approach, including haematologist, paediatric endocrinologist, gynaecologist is the best model for the management of these adolescents.
Methods: We followed 16 girls(13,8±1,2years) with coagulopathies (9 Von Willebrand disease(VWD), 3 inherited platelet dysfunction, 1 Haemophilia A: an extremely rare homozygous mutation in a female; 2 VII factor deficiency, 1 congenital afibrinogenemia) admitted for a metrorrhagic event related to menarche; we evaluated growth (SDS for stature, weight, growth velocity, bone age), pubertal stage, endocrine assess (FSH, LH, PRL, E2), pelvic scan (uterine and ovary morphology and volume; endometrial thickness), haemoglobin, coagulation, coagulation factors concentration, platelet function and aggregation. FSH, LH, PRL, E2, TSH, fT3, fT4, bone age were normal for age. To prevent severe menstrual bleeding the patients received: type I VWD: DDAVP; type II or III: VW and VIII factors (tranexamic acid in all); congenital afibrinogenemia: fibrinogen; Haemophilia A and factor VII deficiency: factor VIII and VII respectively. In girls with concluded PHV and bone age≥14 years, estroprogestinic treatment (E2: 0,03mg-chlormadinone acetate: 2mg) was added.
Results: We observed a significant reduction of menstrual bleeding and a significant reduction in the doses of the specific factors. All the patients were evaluated by DEXA to precociously evidence a reduction in bone mineral density (BMD). BMD values were in the normal range for age and sex, also in patients treated with DDAVP and E2. However our patients received DDAVP for few days/month and E2 from<3years.
Conclusions: We stress the role of endocrine follow up in patients with severe coagulopathies. The correct age to start estroprogestinic therapy must be evaluated by the endocrinologist, to prevent a poor growth prognosis and osteopenia.

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Glucocorticoid resistance (GR) is a novel reason for polycystic ovarian syndrome

Steven Ghanny[1]; Lina Nie[1]; Duanjun Tan[2]; Iuliana Predescu[1]; Natia Pantsulaian[1]; Pascal Philibert[3]; George Chrousos[4]; Constantine Stratakis[5]; Josef Michl[6]; Felicitas Lacbawan[2]; Charles Sultan[3]; Amrit Bhangoo[1]; Svetlana Ten[1]
[1]Infants and Children's Hospital at Maimonides and SUNY Downstate, Pediatric Endocrine, Brooklyn, United States; [2]SUNY Downstate, Molecular Pathology, Brooklyn, United States; [3]CHU de Montpellier, Service d'Hormonologie and Unite d'Endocrinologie Pediatrique, Montpellier, United States; [4]Athens University Medical School, Pediatrics, Athens, Greece; [5]NICHD, Endocrinology and Genetics, Bethesda, United States; [6]SUNY Downstate, Pathology, Brooklyn, United States

Background: PCOS is a heterogeneous group of diseases presenting with ovarian and/or adrenal hyperandrogenism. Although insulin resistance at the level of the ovaries seems to be the main cause of PCOS, other causes have been attributed to the cause of PCOS. There have only been a few reports of glucocorticoid resistance(GR) and hypersensitivity causing PCOS. We present 10 subjects with PCOS who had in addition to elevated androgens, fluctuating elevations of ACTH and/or cortisol levels.
Objective and hypotheses: To study glucocorticoid sensitivity in patients with PCOS.
Methods: We evaluated 10 patients with PCOS(7 overweight, 3 lean) and 15 healthy controls with normal ACTH and cortisol. ACTH stimulation testing normal. 21 hydroxylase mutations were excluded in all patients. F-Dex binding assays were used to evaluate differential binding to the glucocorticoid receptor versus control. DNA was extracted and the glucocorticoid receptor gene(NR3C1), FKBP4 and FKBP5(molecules in glucocorticoid receptor complex) were amplified using PCR and sequence analysis was performed.
Results: F-Dex binding studies in all patients were positive demonstrating 10-50% decrease in binding.
Conclusions: GR has not been shown to be a frequent cause of PCOS. However, screening of our patients with PCOS with fluctuating elevated ACTH and/or cortisol showed 10 patients with decreased F-Dex binding, demonstrating that GR can be a cause of PCOS in these patients.