newborns, particularly in those with at least one other risk factor for HH such as SGA. More studies are needed to elucidate underlying pathology and tease out actual incidence of hypoglycemia in neonates with SARS-CoV-2 infection.

Pediatric Endocrinology

PEDIATRIC ENDOCRINOLOGY CASE REPORT

A Novel Mutation of the AR Androgen Receptor Gene

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Androgen insensitivity syndrome (AIS), formerly known as testicular feminization, is an X-linked recessive condition resulting in a failure of normal masculinization of the external genitalia in chromosomally male individuals. The basic etiology of androgen insensitivity syndrome is a loss-of-function mutation in the androgen receptor (AR) gene. Loss of AR function means that, despite normal levels of androgen synthesis, the typical postreceptor events that mediate the effects of hormones on tissues do not occur. This results in the phenotype of prenatal undervirilization of external genitalia, absence of pubic and axillary hair, lack of acne, and absence of voice changes at puberty We present This baby reffered at age of 2 months from pediatric surgery as a case of bilateral inguinal hernia and chromosomes 46xy. Phenotypically female no male structures no phallus and single opening and visible labia, both test are in the inguinal canals. HCG stimulation test shows: Testosterone: the level at (0) time: 0.8 nmol/l then 3 days: 31.4 nmol/l. DHT dihydrotetostrerone: the level at (0) time: 13 NG/L then 3 days: 485 The baby was given 3 doses of Testosterone injections 150mg but no response at the genetalia. Radiological investigations shows ultrasound both tests at the inguinal canals and no uterus Also MRI of pelvis shows absence of uterus and both test at the inguinal canals Molecular genetics analysis for Androgen receptor gene: Exon 7 c.2512 G >A hem. P.Glu838lys missense, novel VUS,likely pathogenic. The segregations analysis test in the process. Her we present a novel mutation of the AR gene not reported yet in literature.

Pediatric Endocrinology

PEDIATRIC ENDOCRINOLOGY CASE REPORT

A Novel Variant in the CASR Gene c.368T>Cp. (Leu123Ser) in a Case of Hypocalcemia Refractory to Standard Medical Therapy

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Introduction: Hypoparathyroidism is characterized by low or inappropriately normal PTH production, hypocalcemia and hyperphosphatemia. Autosomal dominant hypocalcemia (ADH) type 1 is one of the genetic etiologies of this, caused by heterozygous activating mutations in the *CASR* gene. Some individuals fail to meet treatment goals despite standard therapy. **Clinical Case:** A 13-year-old male patient was admitted in the emergency department due to syncope during physical activity. There was no reference to seizures or other complaints. Standard screening for metabolic diseases revealed no changes at the 7th day of life and family history was unremarkable. There was a history of febrile seizures up to 5 years of age with several hospitalizations for diagnosis investigation that were inconclusive. Physical examination showed a positive Chvostek signal, without other changes. A basic workup revealed hypocalcemia 1.67mmol/L (NR: 2.19-2.66), hyperphosphatemia 3.06mmol/L (NR: 0.95-1.75), hypomagnesemia 0.62mmol/L (NR: 0.7-1.0), low 25Hydroxyvitamin D 8.7ng/mL (NR: >30ng/mL) and inappropriately low PTH 4.0pg/mL (NR: 16.0-87.0). Cranial computed tomography scan showed bilateral calcifications of the basal ganglia. Dual-energy x-ray absorptiometry revealed bone mineral density z-scores increased 15% in spine lumbar and decreased 7% in left femur. Cardiac ultrasound and electrocardiography were normal. The patient started therapy with intravenous calcium gluconate. During this treatment, he developed significant calcification of the peripheral veins at the site of administration, leading to intravenous therapy suspension. The dose of oral calcium, calcitriol and magnesium was gradually increased and sevelamer started to control hyperphosphatemia. Despite the optimization, the patient maintained hypocalcemia refractory to standard therapy. As a last resource strategy in therapeutic optimization, the patient started on rhPTH (1-34). Ever since, the patient has been clinically asymptomatic with biochemical stability and with a reasonable quality of life. At age 18, renal ultrasound revealed diffuse medullary nephrocalcinosis.
The genetic testing revealed a novel variant c.368T>C p.(Leu123Ser) likely pathogenic in heterozygosity in the *CASR* gene, suggesting the diagnosis of ADH type 1. The patient´s mother did not give her consent for genetic study and patient's father had already died with a diagnosis of acute leukemia. **Conclusion:** This case can be explained by the presence of a likely pathogenic variant in heterozygosity in the *CASR* gene that has been described in the medical literature that has not been identified in gnomAD population database, suggesting the diagnosis of ADH type 1. rhPTH (1-34) may be a treatment option for those individuals who are not well controlled on standard therapy, but long-term follow-up studies are needed to reinforce its safety.

Pediatric Endocrinology

PEDIATRIC ENDOCRINOLOGY CASE REPORT

A Pediatric Patient With Noonan Syndrome and Late Onset Unilateral Lymphedema

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Introduction: Noonan syndrome is a common autosomal dominant disorder with a prevalence of 1 in 1000-2500 births. The lymphatic disorders in Noonan syndrome are rare and usually bilateral. We present a 14-year-old male