of 45, X accounts for nearly 50% of patients, while mosaicism and other chromosomal structural abnormalities such as deletions, duplications, ring, isodicentric chromosomes, inversions and translocations, have been reported. Isodicentric X chromosomes are formed presumably by end-to-end fusion of chromatids after a break, with subsequent loss of an acentric fragment. These chromosomes in general have phenotypes characteristic of the resultant X deletions. We present a case of a 14-year-old female diagnosed with Turner syndrome and with 2 abnormal cell lines. **Case Presentation:** This is a case of a 14-year-old female referred to pediatric endocrinology for concerns of short stature and delayed puberty. She denied any food intolerance, bloating and diarrhea. She is otherwise healthy with unremarkable past medical history. Her weight was normal at 15th percentile. Her height was 137cm or 0.01 percentile with a Z score of –3.6. Work up revealed hypothyroidism with TSH 16.3 mcIU/mL (0.4-4.7 mcIU/mL), positive thyroid peroxidase antibody >900 IU/ml and thyroglobulin antibody 14 IU/mL (< 1.8IUm/mL) and celiac disease (tissue transglutaminase IgA > 100 U/mL) both without associated symptoms. Estradiol level was undetectable, and LH and FSH were 9.89 mIU/ml and 52.69 mIU/ml respectively. The rest of her labs including growth factors were normal. Bone age was normal at 13 years for chronological age of 14 years old. Chromosomal microarray revealed 2 abnormal cell lines: one with monosomy X, the other with a normal X chromosome and an isodicentric X chromosome involving the Xp11.22-q28 region resulting in trisomy of the latter cell line.

Levothyroxine was started. Plan is to start growth hormone therapy and initiate puberty after. Patient referred to necessary subspecialties for hearing evaluation as well as cardiac evaluation Conclusion

Turner syndrome usually presents as females with short stature, gonadal dysgenesis and 45,X cell line that is either singly or in combination with another

mosaic cell line. Our patient presented with short stature and absence of puberty. Initial investigation revealed hypothyroidism and highly positive celiac antibodies, but unable to attribute her short stature to both diagnoses given the lack of other symptoms. This case emphasizes the importance of checking the karyotype in females presenting with short stature and more importantly delayed puberty as part of the diagnostic algorithm. In addition, checking thyroid and celiac panel are also imperative as treatment of these are treatable etiologies of short stature.

Pediatric Endocrinology

PEDIATRIC ENDOCRINOLOGY CASE REPORT

Approaching High-Metabolic Risk Youth During a Pandemic: Severe Presentations of New Onset Type 2 Diabetes

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Background: Delays in timely medical care due to the restrictions imposed by the COVID-19 pandemic have worsened patient outcomes with different disease conditions. Youth with obesity, insulin resistance, and dysglycemia are increasingly presenting with HHS and/or DKA, leading to increased morbidity and mortality. **Case Descriptions: Case 1**: A 17-year-old Hispanic female with history of obesity, insulin resistance, and hyperglycemia reported polyuria, polydipsia, and polyphagia for 2 months. The day of admission, EMS was called because patient was found unresponsive at home and required advanced CPR. At arrival to the ED, the patient was awake and responsive. She had acanthosis nigricans and abdominal striae. Initial labs showed elevated plasma glucose (1,256 mg/dL), sodium (153 mmol/L), bicarbonate (9 mmol/L), anion gap (35 mmol/L), phosphorus (7.5 mg/dL; N=3-4.8), lactate (4 mmol/L; N=0.5-2), BHB (11.4 mmol/L; N=0-0.3), venous pH (7.09), BUN (24 mg/dL), creatinine (1.63 mg/dL), and HbA1c (14.3%). She was admitted for DKA, hyperosmolarity, AKI, and metabolic encephalopathy. After IVF resuscitation, insulin drip at 0.05 U/kg/h was started. She recovered from DKA, AKI and hyperosmolarity after 5 days. T1D antibody tests were negative. C-peptide was low (0.7 ng/mL; N=0.8-3.5), and TSH was low (0.38 uIU/mL) with normal free T4 (0.88 mg/dL). **Case 2**: A 13-year-old Hispanic female with history of asthma, morbid obesity, premature adrenarche, and prediabetes started presenting polydipsia and polyuria 2 months before admission. One day before admission, she presented drowsiness, abdominal pain, and polyuria. Initial labs at the ED included glucose (792 mg/dL), bicarbonate (10.4 mmol/L), anion gap (28 mmol/L), venous CO_2 (10.4 mmol/L; N=21-31), BHB (>22.5 mmol/L), sodium (153 mmol/L), BUN (29 mg/dL), and creatinine (1.37 mg/dL), consistent with DKA, hyperosmolarity, and AKI. Physical exam showed severe obesity, acanthosis nigricans, and hypertension. IV fluids and insulin drip at 0.075 U/kg/h were started. DKA and AKI resolved after 4 days. Labs showed negative T1D antibodies, normal C-peptide (1.2 ng/mL), HgA1C (>14%), microalbuminuria (16.22 mg/dL; N<2 mg/dL), elevated total cholesterol (230 mg/dL) and triglycerides (550 mg/dL). Both youth were negative for SARS-CoV2 and had been engaging in unhealthy lifestyle choices, such as sedentarism and excessive sugary drink intake, exacerbated by COVID-19-related lockdowns and school closures. There were delays in seeking medical care associated to fear of COVID-19. **Conclusion**: In the current context of a pandemic, it would be helpful to plan close evaluation and timely therapeutic interventions for youth with well-known high-metabolic risks to prevent hospitalizations, severe presentations of T2D and associated morbidity and/or mortality.

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PEDIATRIC ENDOCRINOLOGY CASE REPORT

Case Report and Literature Review: Homozygous DNAJC3 Mutation in a Saudi Family Causing Maturity Onset Diabetes of the Young (MODY), Hypothyroidism, Short Stature, Neurodegeneration, and Hearing Loss

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Introduction: Monogenic diabetes results from a mutation in single gene, predominantly inherited and typically affects the young. DNAJC3 acts in attenuating endoplasmic reticulum stress and is found in abundance in pancreatic tissue. **Clinical Case:** We report a homozygous DNAJC3 mutation in two siblings of a consanguineous Saudi family. A 3-year boy presented with short stature and thyroid nodule; lab findings confirmed hypothyroidism, with TSH 27.8 and FT4 6.7 (n: TSH:0.35-4.94 mIU/L, FT4:9.0-19 pmol/L). Subsequently, L-thyroxine was started. GH stimulation test was normal. He was severely short; 80.5 cm (< 1 percentile, -3.79 SD). The patient developed sensorineural hearing loss (SNHL) at 6 years. He had low intellectual function and weak school performance. GH treatment was postponed to age 9 due to strong family history of DM. At that point, the patient developed progressive ataxic gait, for which he had muscle biopsy that excluded mitochondrial disease and workup for multiple sclerosis, which was excluded. Brain and spine MRI showed prominent neurodegeneration in subcortical white matter. At age 11, the patient developed DM, 4 years after GH treatment initiation. DM autoimmune markers were negative on multiple occasions. Lifestyle modification was initiated but soon required basal and bolus insulin therapy. Whole exome sequencing revealed homozygous DNAJC3 mutation, which explained his clinical presentation. At age of 17, adult height was 141 cm (Z-score: -5.87). His older brother had similar history discovered retrospectively but did not develop neurodegeneration or ataxia from the same DNAJC3 mutation. **Literature Review:** Literature review revealed six individuals with homozygous DNAJC3 mutation. All patients developed DM, with onset ranging from 11 to 19 years, highly suggestive of MODY. Other endocrine manifestations included short stature, and hypothyroidism due to primary etiology; in view of elevated TSH levels, vs. being secondary, as suggested by the authors. All patients had mitochondrial disease workups and was excluded. Variable neurodegeneration degrees are described; SNHL, progressive ataxia, sensorimotor neuropathy, and cognitive deficits. MRI findings showed atrophy of cerebellum, brainstem, cervical spinal cord, and hyperintense T2 lesions typical of neurodegeneration. **Conclusion:** Homozygous DNAJC3 gene mutation fits MODY criteria, we propose recognizing it as one of the known MODY gene mutations. Hypothyroidism is due to primary etiology, evident by TSH spikes. Physicians evaluating mitochondrial disease in patients with a constellation of SNHL, DM, hypothyroidism, neurodegeneration, and short stature should suspect DNAJC3 as one differential diagnosis. GH treatment must be initiated cautiously, with close monitoring due to its known diabetogenic effect, especially in DNAJC3 mutations, defective endoplasmic stress attenuation mechanism.

Pediatric Endocrinology

PEDIATRIC ENDOCRINOLOGY CASE REPORT

Case Report: Primary Hypothyroidism Caused by Autoimmune Thyroiditis in Infancy Requires Early Intervention

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Background: Primary hypothyroidism due to autoimmune thyroiditis is extremely rare in infants, especially under the age of 3 years. For infants, hypothyroidism is most commonly congenital, originating from thyroid dysgenesis with an absent, ectopic, or hypoplastic thyroid gland (1 in 4,000 live births). If left untreated, it can lead to permanent neurodevelopmental deficits. In this report, we describe a male infant who was diagnosed with Hashimoto thyroiditis at 18 months of life, providing a learning example to aid in recognition of this rare disease and enable timely intervention.

Clinical Case:Patient was a 2,765 gram, appropriate for gestational age, male born at term with hypospadias of the penis (surgical correction at 11 months). Patient passed meconium in the first 24 hours of life. During the first few months of life, patient developed constipation. Patient had amblyopia necessitating eye patching and began to wear eye glasses at 18 months of life. Patient's linear and weight growth were within normal limits. Patient had normal motor development, however had language development delay. No known family history of thyroid disease. Screening labs performed at 17-months of age showed abnormal thyroid function: elevated TSH at 14.86 μIU/mL (ref: 0.45 - 4.50 μIU/mL) and normal free T4 level at 1.24 ng/dL (ref: 0.85-1.75 ng/dL). Repeat testing at 18 months of age continued to show elevated TSH at 6.18 μIU/mL (ref: 0.64 - 4.00 μIU/mL), normal free T4 at 1.07 ng/dL (ref: 0.88 - 2.03 ng/dL), and elevated thyroid peroxidase (TPO) antibodies at 163 IU/ml (ref: <35 IU/ml). At 21 months of age, patient was initiated on L-thyroxine therapy for elevation of TSH (9.570 μIU/mL; ref: 0.64 - 4.00 μIU/mL) and free T4 was normal (1.03ng/dL; ref: 0.88 - 2.03 ng/dL). Notably, the newborn screen for hypothyroidism was within normal limits, suggesting chronic autoimmune thyroiditis instead of congenital thyroid dysgenesis.

Conclusions: This case report provides insights into autoimmune thyroiditis in infancy, which, although especially rare under age 3 years, should be considered in infants who present with autoimmunity or abnormal thyroid testing. In the neonatal period, infants' immune systems are learning to discriminate requirements for self-tolerance versus protection against pathogens and may be more prone to infections. Although autoimmunity in this stage of development is uncommon, there can be breakthroughs in tolerance, as seen in this case. In addition to this patient, two other infants were seen with elevated TPO antibodies, diagnosed at 17 and 31 months old, with similar clinical trends. There remains a need for additional studies providing further insights into autoimmunity in infancy. Importantly, this case illustrates that, when infants have abnormal thyroid levels (with or without other autoimmune