

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 (< 1.8IU/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.

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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 CO₂ (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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