

family renamed the child “Brenda.” Unaware of her history, Brenda struggled with significant gender identity, psychological, and behavioral issues throughout her childhood and adolescence. When made aware of this history, she transitioned to male gender and assumed the name “David.” After years of psychological distress, David Reimer committed suicide in 2004. Despite the myriad lessons gleaned from this tragic story, medical and surgical management of children with atypical genitalia still remains often misguided, as providers continue to assume paternalistic roles in determining sex assignment and surgical interventions. A fifteen year old XY male with Robinow Syndrome presented for evaluation of hypogonadism and urinary incontinence. At birth, the patient was discovered to have a micropenis and perineal hypospadias and was diagnosed with hypogonadotropic hypogonadism. At the recommendation of the medical team, the infant underwent bilateral orchiectomy at eight months of age followed by urethroplasty and vaginoplasty at six years of age. The child was then given a female sex assignment. At twelve years of age, the child felt discordant from the sex of rearing and wished to be identified as male—his natal, genetic sex. He transitioned to male gender and began testosterone injections. He had history of recurrent UTIs and severe incontinence requiring diaper use. He strongly desired neophallus and urethral reconstruction for improved quality of life. The patient endorsed prior depression and desires to self-harm. He had significant concerns regarding his gender presentation and transition. He shared his difficulties in continuing in the same school system with peers who knew him as a female prior to transition and was concerned about peers knowing his medical history. In the years since the famous David Reimer case, the medical system has made tremendous strides in recognizing the need for patient autonomy and shared decision-making in patients with Differences of Sex Development and genital atypia. However, the paternalistic history of this field continues to leave its indelible mark more than 20 years since David Reimer’s case made headlines, as physicians continue to recommend definitive sex assignments and surgical interventions. As with the David Reimer case, the bodily integrity of this XY infant was altered in a permanent fashion with inadequate education of his family and little to no credence given to the autonomy of the child himself. We, as physicians, cannot continue to paternalistically apply John Money’s concept of gender neutrality and rigidly mandate sex assignments and early surgical interventions.

## Pediatric Endocrinology

### PEDIATRIC ENDOCRINOLOGY CASE REPORT

#### *Pediatric Giant Prolactinoma Presenting With Acute Obstructive Hydrocephalus and Intracranial Hypertension*

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**Background:** Pediatric prolactinomas (PP) are rare but represent 50% of all pediatric pituitary adenomas. Girls are affected more frequently than boys, although PP tend to be larger and more aggressive (earlier age, larger mass, and higher prolactin levels) in boys. Thus, microadenomas (tumors < 10 mm in diameter) are typical in females and macroadenomas (10–40 mm in diameter) are typical in males. Giant prolactinomas (> 40 mm in maximum diameter), an unusual subset of macroprolactinomas, are also commonly found in boys. In a large case series, the largest tumor volume reported was 93.5 cm<sup>3</sup>. Here we report a giant prolactinoma in a female requiring V/P shunt for decompression. **Clinical Case:** A 16-year old female presented with 2 weeks of intractable headache, nausea and vomiting, vision impairment, and changes in balance described as running into stationary household objects. Historical review revealed primary amenorrhea and short stature. On initial exam, the patient had a right eye afferent pupillary defect, concern for loss of color vision, and bilateral optic nerve edema with blurred disc margins. Brain MRI showed a large lobulated mass centered in the suprasellar cistern, measuring approximately 6.4 x 5.8 x 5.7 cm with a tumor volume of 105 cm<sup>3</sup>. There was extension superiorly, anteriorly, and laterally, with homogeneously enhancing and cystic components, and mass effect resulting in obstructive hydrocephalus. Differential diagnoses included craniopharyngioma, germinoma, and adenoma. Initial tests demonstrated prolactin of >2,000 ng/mL, with diluted result of 17,811.16 ng/mL. Morning fasting labs confirmed multiple anterior pituitary hormone deficiencies including central hypothyroidism, ACTH deficiency, GH deficiency, and hypogonadotropic hypogonadism. The patient was started on hydrocortisone and levothyroxine. Due to obstructive hydrocephalus and vision impairment, she underwent VP shunt placement for decompression. She was started on cabergoline for medical treatment of the tumor and did not require surgical resection. Repeat prolactin measurements have shown striking improvement (to 2,350 ng/ml, 824 ng/ml, and 152 ng/ml at 1 week, 1-month, and 2-month-follow-up, respectively) with central vision improved in both eyes, papilledema resolved, and resolution of headaches. **Conclusion:** Giant prolactinomas presenting with hydrocephalus and intracranial hypertension are very rare in pediatrics, especially in girls, and can vary greatly in mass characteristics and resulting hormone deficiencies. Our patient is unique with her large tumor volume and the extent of pituitary hormone deficiencies. Prolactin levels should be measured with all sellar masses, as this may prevent unnecessary invasive intervention and possibly provide prompt response to medical management.

## Pediatric Endocrinology

### PEDIATRIC ENDOCRINOLOGY CASE REPORT

#### *Pituitary Macroprolactinoma Apoplexy in a Prepubertal Girl*

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**Background:** Prolactinomas are rare in children, with an incidence of 1: 10,000,000 cases, representing less than 2% of all intracranial tumors in this age group (1). Indeed, only a few cases are reported in prepubertal children.

**Clinical Case:** Eight-year old female prepubertal child, previously healthy, presented a progressive loss of the visual acuity in the last 5 months and was admitted to the emergency department with an intensive headache associated with nausea, started 48 hours before the admission. At the physical examination, the patient was eutrophic, with an infantile genitalia and no breast development or galactorrhea. No focal neurologic deficits were detected and presented normal pupils reflex and preserved extrinsic eye movements. The Campimetry evidenced bitemporal heteronymous hemianopsia and the nuclear magnetic resonance of the central nervous system identified a suprasellar and intrasellar expansive lesion measuring 3.2 x 2.6 x 2.3 cm, with bleeding signs and compression of the optic chiasm. The hormonal evaluation showed: free T4 0.55 ng/dL (normal value: 0.93-1.70ng/dL), total T4 4.8ng/dL (5.1-14.1ng/dL), TSH 4.06 $\mu$ UI/mL (0.6-5.4 $\mu$ UI/mL), morning serum cortisol 1.80mcg/dL (6.2-2mcg/dL), ACTH 7.0pg/mL (<46pg/mL), prolactin 3.376ng/mL (4.8-23.3ng/mL). The patient initially started glucocorticoid replacement, subsequently levothyroxine and cabergoline. A diagnosis of macroprolactinoma with pituitary apoplexy and optic nerve compression was performed and confirmed by the Immunohistochemical after the transsphenoidal resection. No history of any neoplasms were reported by family members and calcium abnormalities were excluded in the child. A significant improvement of the visual field was suddenly observed and the prolactin substantially decreased to 35ng/mL within 30 days after the intervention. **CONCLUSION:** We report a rare case of macroprolactinoma in a prepubertal girl associated with pituitary apoplexy. Complete assessment and prompt intervention were essential to recover the visual field and to prevent major sequelae.

**Reference:** (1) Yang, A., Cho, S. Y., Park, H., Kim, M. S., Kong, D.S., Shin, H.J., & Jin, D.K. (2020). Clinical, Hormonal, and Neuroradiological Characteristics and Therapeutic Outcomes of Prolactinomas in Children and Adolescents at a Single Center. *Frontiers in Endocrinology*. 2020; 11:527.

## Pediatric Endocrinology

### PEDIATRIC ENDOCRINOLOGY CASE REPORT

#### *Primary Amenorrhea and Hyperandrogenism: Presenting Features of a Growth Hormone Producing Pituitary Adenoma in a Female Adolescent*

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**Background:** The combination of obesity, metabolic syndrome, hyperandrogenism and amenorrhea can be a common presentation of conditions such as polycystic ovarian syndrome. Growth hormone (GH) secreting pituitary adenomas are rare in children, but can present with a similar picture. **Clinical Case:** A 15-year 4-month old

female was evaluated for primary amenorrhea and metabolic syndrome. Her weight was +2.96 SD, height +3.0 SD (mid-parental height 44<sup>th</sup> percentile, -0.1 SD) and BMI +2.53 SD, all >99<sup>th</sup> percentile. Weight and BMI were >99<sup>th</sup> percentile (weight +3.0 SD, BMI +2.4 SD) since 11 years of age and height was >99<sup>th</sup> percentile (+ 2.7 SD) since 8 years of age. Physical exam was remarkable for acanthosis, deepened voice, no hirsutism, Tanner III breasts and Tanner IV pubic hair. Laboratory evaluation was notable for prolactin (PRL) 147.8 ng/dL (2.6-18.0), LH <0.02 mIU/mL, FSH <0.09 mIU/mL, estradiol 23 pg/mL, total testosterone 81 ng/dL (<41), androstenedione 673 ng/mL (43-180), DHEA-S 627 mcg/dL (42-162), dyslipidemia and normal 17 hydroxy-progesterone and thyroid function tests. Bone age was 16 years. Brain MRI showed a 15x7mm pituitary macroadenoma without suprasellar invasion. Pituitary evaluation revealed IGF-1 619 ng/mL (192-568), IGF-BP3 5.15 mg/L (2.64-6.43) and adrenal insufficiency. Treatment with cabergoline 0.5 mg/week and maintenance hydrocortisone were initiated. An OGTT demonstrated baseline GH 4.9 ng/mL (0-6) and nadir 3.62 ng/mL (normal <1) consistent with GH excess. Surgery for the pituitary adenoma was recommended but unfortunately postponed. Four months later, her BP was elevated, she had clinical signs of diabetes insipidus (DI), PRL was 43.2 ng/dL and she failed a repeat GH suppression test. Follow-up MRI showed pituitary mass enlargement (17x9mm). Patient underwent total trans-sphenoidal tumor resection. Pathology confirmed a sparsely granulated (SG) GH adenoma immunoreactive for GH, PRL and estrogen receptor. Post-operative OGTT showed adequate GH suppression, normal prolactin levels, persistent testosterone elevation and DI. **Conclusions:** Hyperprolactinemia, hyperandrogenism and metabolic syndrome could be the presenting features of pituitary GH adenomas even in the absence of acromegalic features. Specifically, SG pituitary adenomas are frequently seen in young adults, are larger, more invasive and less likely to respond to medical treatment. Timely diagnosis and surgical treatment of GH adenomas is essential to prevent the high morbidity and mortality.

## Pediatric Endocrinology

### PEDIATRIC ENDOCRINOLOGY CASE REPORT

#### *Steroid Cell NOS in a Child: A Rare Case Report*

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**Introduction:** Steroid cell tumors are rare sex chord tumours of the ovary with malignant potential. Here we report a case of steroid cell NOS in a 12-year-old girl & similarity of the clinical presentation with non-classic congenital adrenal hyperplasia.

**Clinical Case:** 12-yr girl came with complaints of hirsutism since many years and a recent change in voice. There was no past history suggestive of adrenal crisis. On examination she had muscular body habitus, moderate hirsutism (FG score 19), acne. SMR B1, P4, A3. Karyotype 46 XX. BA 13yrs. DHEAS- 144, serum cortisol 8am-14ug/dl. Testosterone-8.35ng/ml (normal 0.1-0.9ng/ml). serum 17 (OH) Progesterone basal-16.37 ng/ml(N-0.2-1.3ng/ml). Post synacthen 17OHP >35ng/ml, LH- <0.1,