Initial work-up with peripheral blood, iron studies and haemoglobin analysis confirmed true thrombocytopenia and ruled out haemoglobinopathies. Screening of the anterior pituitary hormone profile revealed that she had hypothyroidism, hypocortisolism, growth hormone deficiency, low gonadotropins levels with normal prolactin. MRI demonstrated a small anterior pituitary measuring 0.4 cm, with an ectopic posterior lobe and the infundibulum was not visualised. Her presentation, complemented with the biochemical and radiological findings confirmed a diagnosis of PSIS. She was started on hormonal replacement therapy.

CONCLUSION

PSIS occurs in about 0.5/100,000 births. The pathogenesis is largely unknown, though genetic factors and obstetric trauma are considered potential contributors. Common presentation includes short stature and delayed puberty. A combination of clinical assessment and biochemical tests is required to form a suspicion with MRI as the confirmatory test, differentiating PSIS from other pituitary pathologies. Treatment involves replacing the deficient hormones.

EP_A117

A RARE CASE OF KALLMAN SYNDROME IN A FEMALE

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INTRODUCTION/BACKGROUND

Kallman Syndrome, a rare genetic disorder referring to hypogonadotropic hypogonadism, is associated with anosmia or hyposmia. It is due to abnormal migration of gonadotrophin-releasing hormone-producing neurons. The reported incidence is 1 in 8000 in men and is 5 times rarer in women. We reviewed case notes, investigation results, and imaging studies and discussed treatment options based on literature review and treatment availability.

CASE

A 19-year-old female was referred to endocrinology for primary amenorrhoea with pituitary microadenomas. The patient was born to non-consanguineous parents. She is the second child with 3 healthy siblings. The patient cannot smell since childhood. On physical examination, Tanner's staging of the breasts and pubic hair were 3 and 1, respectively, with no axillary hair. Perineal examination revealed a not well-formed labia majora with well-formed labia minora. Urethra and vaginal orifice were seen. She had bilaterally small fingers. Ultrasound of the abdomen showed a small uterus at 2.1 x 1.1 cm with no ovaries seen. MRI of the brain showed bilateral pituitary microadenomas measuring 5 x 4 x 3 mm and 5 x 4 x 2 mm on the right and left side of the anterior pituitary lobe, respectively. Unfortunately, the olfactory bulb was not assessed. Hormonal assays identified a hypogonadotropic hypogonadism profile with total serum testosterone <0.24 nmol/L (NR: 0.29-1.21 nmol/L), serum oestrogen <43.3 pmol/L (NR: 59.1-874.6 pmol/L), serum luteinizing hormone 0.11 IU/L (NR: 1.0-52.5 IU/L), serum follicular stimulating hormone 1.02 IU/L (NR: 2.2-10.1 IU/L). Serum prolactin was normal at 79.23 uIU/mL. The patient was started on oestrogen pills and started to have fullness and tenderness in her breasts. She was referred to the Genetic Clinic for genetic studies.

CONCLUSION

Patients presenting with primary amenorrhoea and anosmia should prompt suspicion of Kallman Syndrome. Laboratory and radiological evaluation may be helpful as genetics confirmation will take time. Early detection and initiation of hormonal treatment will enable the progression of the secondary sexual characteristics. However, achieving fertility will still be a challenge depending on the availability of gonadotrophins or pulsatile GnRH therapy.

EP_A118

AN AGGRESSIVE CATECHOLAMINE-SECRETING GLOMUS PARAGANGLIOMA: A CASE REPORT

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INTRODUCTION/BACKGROUND

Head and neck paragangliomas (HNPGLs) are commonly detected at the carotid artery bifurcation and carotid body but may arise in the middle ear. HNPGL presents as slow-growing, painless neck mass. Majority are non-functional with approximately 5% being biochemically active. HNPGLs are generally locally invasive, and destructive and up to 19% may be malignant. Management for this type of paraganglioma is difficult and requires a multidisciplinary approach. We present a patient with a huge and locally aggressive functioning left glomus-jugulotympanicum-paraganglioma with significant management challenges.

CASE

A 33-year-old male presented with left ear pulsatile tinnitus which was treated as left otitis media. Despite the persistent symptoms and progressive hearing

impairment in the left ear, an investigation for suspicion of glomus tympanicum commenced 6 months later. He had repeated ER visits for the next year due to the persistence of symptoms and uncontrolled hypertension. However, he was only referred to the endocrine clinic for young hypertension investigation 20 months from the initial presentation with significant paroxysms of palpitation, headaches and elevated BP. Diagnosis of catecholamine-secreting glomus-jugulotympanicum-paraganglioma was confirmed with elevated urinary metanephrine and huge soft tissue mass in the left jugular fossa with local bony erosion and intracranial extension on MRI.

His BP control was labile and required multiple oral-antihypertensives including phenoxybenzamine. Multi-disciplinary team management prepared him for definitive surgical intervention. He underwent tumour embolization prior to the actual surgery. Pre-operative management was extremely challenging which required CCU admission for BP stabilization. Intra-operative period was surprisingly uneventful, but he developed multiple cranial nerve palsies postoperatively. A second operation was required due to infection and enlarging tumor with compression. Paroxysm of symptoms improved after second surgery but he still had significant residual tumor. MIBG-therapy was planned but management was delayed due to the COVID-19 pandemic and treatment funding.

CONCLUSION

Awareness of functional paraganglioma presentation is imperative to avoid late detection. HNPGLs that are both aggressive and functional pose extreme difficulty in achieving disease remission.

EP_A119

ADIPSIC CENTRAL DIABETES INSIPIDUS IN A PATIENT WITH SUPRASELLAR GERMINOMA

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INTRODUCTION/BACKGROUND

Adipsic central diabetes insipidus (CDI) is a rare and life-threatening disease which is difficult to manage as the patient experiences loss of thirst sensation compounded by fluid loss and hypernatremia. We present a case of a patient with suprasellar germinoma with panhypopituitarism who presented with adipsic CDI.

CASE

A 20-year-old male presented with generalized body lethargy for 1 month and polyuria for 1 year. He did not complain of polydipsia. Blood tests revealed severe hypernatremia with serum sodium (Na) of 160 mmol/L, serum osmolality of 300 mOsm/kg and urine osmolality of 130 mOsm/kg. Other electrolytes and blood glucose were normal. MRI showed a large sellar/suprasellar mass with periventricular subependymal spread causing acute obstructive hydrocephalus. Hormonal panel showed panhypopituitarism. He was started on sublingual desmopressin, L-thyroxine, hydrocortisone and intramuscular testosterone. During his confinement, he denied polydipsia despite intermittent polyuria. Strict intake and output monitoring were instituted with hourly urine output and regular renal profile monitoring. Despite initial normalisation of Na levels, he developed 2 episodes of hypernatremia when he had breakthrough polyuria. Intravenous fluids were given intermittently to balance his output. The patient and caregiver were constantly reminded to take adequate oral fluid. He then underwent transcranial biopsy. Histopathology examination showed a diagnosis of central nervous system germinoma. A referral to the Oncology team for chemotherapy was made.

Adipsic CDI has been reported to account for about 10% of CDI cases. Patients with adipsic CDI have higher prevalence of complications such as hypernatremia, renal insufficiency and venous thrombosis. Apart from desmopressin, crucial management steps include regular monitoring to ensure adequate fluids and desmopressin replacement.

CONCLUSION

This case highlights the difficulties in managing adipsic CDI and the need for constant and regular monitoring to prevent life-threatening hypernatremia.