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.

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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