**Introduction:** Two childhood cases of somatotroph pituitary adenomas caused by AIP gene mutations highlight the importance of screening for Familial Isolated Pituitary Adenoma (FIPA) genes in index cases and family members.

**Case 1:** 13.5 year old girl presented with 5 years growth acceleration and size 10 feet. No headaches or visual disturbance.

**Examination:** Acromegalic facies, large hands and feet. Height 177cm (>99.6th centile, SDS +2.8). Mid parental centile +25th centile.

**Biochemistry:** OGTT showed elevated baseline GH with marked failure to suppress and impaired glucose tolerance. IGF-1 208nmol/l (23-90). Pituitary function otherwise normal.

**MR1:** 18mm pituitary mass with suprasellar extension.

**Histology:** Pituitary adenoma (Fig1a). Immunohistochemistry shows adenoma cells strongly positive for GH but largely negative for ACTH, FSH, LH, TSH and prolactin (Fig1b). Overall appearances consistent with GH secreting adenoma.

**Post surgical resection:** Developed sphenoidal abscess resulting in permanent left temporal upper quadrantopia. Subsequently normalisation of IGF-1, growth and normal pituitary function

**Post operative course:** IGF-1 remained elevated (91.4nmol/l [9.8-61.0]) 6 weeks post resection. No preoperative sample for comparison due to insufficient sample being sent.

**OGTT 6 weeks post-op:** Ongoing GH hypersecretion.

**Medical Management:** Octreotide commenced as shown but IGF-1 remained elevated. Patients with AIP mutations are known to respond poorly to somatostatin analogue therapy. Further surgical clearance undertaken. Further therapy options include GH receptor antagonist or pasireotide treatment.

**Genetics:** A novel heterozygous frameshift mutation in AIP c.376_377del:p.Q126fs was identified. Her mother was an asymptomatic carrier (IGF-1 18.8nmol/l [7.0-25.9], prolactin 248mIU/l [<700], suppressed GH on OGTT [nadir 0.2mcg/l]). Her siblings tested negative and mother’s family is undergoing genetic testing.

**Case 2:** 10 year old boy presented with daily headaches and sudden onset blurred vision with recent growth acceleration and increase in shoe size.

**Examination:** Slight coarsening of facies, blurred infero-temporal visual fields. Height SDS +3.05.

**Histology:** Pituitary adenoma immuno-positive for GH, but negative for other pituitary hormones.

**Summary:** Both these cases of childhood onset GH secreting adenomas were index cases of Familial isolated Pituitary Adenoma (FIPA). 50% of identified AIP kindreds have no known family history. Prompt AIP mutational analysis in such cases facilitates management focussed on GH excess and allows discontinuation of other endocrine tumour screening. Family testing is important to allow early identification of GH excess in family members.

Ref: Leoniou et al; The role of the aryl hydrocarbon receptor-interacting protein gene in familial and sporadic pituitary adenomas; J Clin Endocrinol Metab. 2008 Jun;93(6):2390-401.