Hypothalamic hamartomas (HH)- rare heterotopic congenital malformations (incidence 1:200000)- present with central precocious puberty (CPP) or gelastic seizures (GS) but their natural history and best treatment strategy are unknown. Given their proximity to the hypothalamus-pituitary axis, wider endocrine dysfunction may be expected.

**OBJECTIVE AND HYPOTHESES**

To describe clinical features and any evolving endocrinopathies in HH patients, by presentation (CPP or GS), radiological characteristics and any surgical treatment imposed.

**METHODS**


**RESULTS**

- Over a 14 year period, 34 children (21M:13F) aged 3.97 (±3.5 SD) years were diagnosed with HH and followed for 5.9 (±4.3 SD) years.
- Patients’ main clinical and radiological presenting features are shown in Table 1 and Table 2.
- Fourteen (41.2%) patients each, presented with either GS or CPP and a further 6 were identified incidentally (MRI) [Table 1]. Patients presenting with GS tended to be older than those with CPP or incidental HH and all had sessile hamartomas. By contrast, most HH in CPP and incidental groups were pedunculated [Table 2].

- 28.6% patients with GS developed CPP 3.16 (±2.8SD) years later at 4.5 (±3.85D) years old (in 3 after surgery). Similarly, 28.6% CPP patients were diagnosed with GS (2/4 sessile) 0.57 (±0.41SD) years later.
- 32.3% (11/34) patients (10 presenting with GS and 1 with CPP), all with refractory GS, underwent surgery 3 years (±2.6) from diagnosis.
- At last assessment, half of presenting GS patients had pituitary deficits compared with none of CPP and just 16.6% of incidental. Only 4 (all post-operative) had Central Diabetes Insipidus (CDI), 3 with additional GH deficiency (GHd) & TSH deficiency (TSHd).
- Obesity rates were high, especially in those presenting with CPP and GS [Table 3].

**CONCLUSION**

- GS HH and CP HH are an overlapping spectrum.
- CPP HH (especially if sessile) require routine neurology referral (to exclude occult GS).
- GS HH require routine endocrine review, especially if surgery - which is the only significant cause of DI - is contemplated.
- Routine dynamic pituitary testing (including LHRH) of all HH patients may unmask CPP and GHd, whilst prompt GH replacement may reduce the currently high obesity rates.