ENDOCRINE DYSFUNCTION IN HYPOTHALAMIC HAMARTOMA DEPENDS ON PRESENTATION (ENDOCRINE OR EPILEPTOGENIC), RADIOLOGICAL CHARACTERISTICS AND SURGERY

Great Ormond Street Hospital for Children NHS Foundation Trust

Elisabetta Caredda[1], Robert Wilkinson[3], Ash Ederies[4], Nicola Improda[1], Sophia Varadkar[3], Helen Cross[3], Martin Tisdall[2], Helen A Spoudeas[1]

Departmentof Paediatric NeuroEndocrinology[1], Neurosurgery[2], Neurology[3] & Neuroradiology[4],
Great Ormond Street Hospital for Children, London UK

BACKGROUND

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

Retrospective-longitudinal review of case notes of children with HH seen at our centre between 1.1.1991 and 31.12.2014.

RESULTS

CLINICAL PRESENTING FEATURES

Table 1

	CPP	GS	Incidental	P value
N (%)	14 (41.2)	14 (41.2)	6 (17.6)	
Sex M/F (%)	5/9(35,7/**64,3**)	13/1(**92,8**/7,2)	3/3(50/50)	<0.005
Age Diagnosis HH y (SD)	3,12(±2,46)	5,09(±3,8)	3,72(±4,45)	ns

RADIOLOGICAL AND PRESENTING FEATURES

MRI-Grading - Table 2

		CPP	GS	Incidental	P value
MRI Type	Sessile (%)	21.4	**100**	16.6	ns CPP-INC
	Pedunculated (%)	**78.6**	0	**83.4**	

FOLLOW UP

Endocrine comorbidities - Table 3

		CPP	GS	Incidental	P value	Total
CPP		14(100%)	4 (28,6%)			18 (53%)
Pituitary deficit	TOT		**7(50%)**	1 (16,7%)	ns	8 (23,5%)
	CDI		4 (28,6%)			4 (11,7%)
	GHd		3 (21.4%)	1 (16,6%)	ns	4 (11,7%)
	TSHd		3 (21.4%)	1 (16,6%)	ns	4 (11,7%)
	MPDH		3(21.4%)	1 (33%)	ns	4 (11,7%)
Obesity (BMI >2SDS)		60%	46%	20%	ns	46,4%
Insulin Insensitivity		1 (7%)	2(14%)	1 (16,6%)	ns	4 (11,7%)
Epilepsy		4 (28.6%)	14(100%)			18 (53%)
Surgery		1 (7.1%)	10 (**71.4%**)	0	<0.005	11(32,3%)

- 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 (±3.8SD) 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 3.3years (±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.

ESPE 2015 1148--P3
Puberty
Elisabetta Caredda
DOI: 10.3252/pso.eu.54espe.2015
Poster presented at: 54th Annual Meeting 1-3 October ESPE 2015 BARCELONA European Society for Paediatric Endocrinology
PosterSessionOnline.com