

Hypothalamic Hamartoma Causing Epilepsy Patient Journey



First Symptom

Laughing/smiling, eye or mouth movements; reflux. The laughter is in fact seizureal – gelastic seizures – and points to hypothalamic hamartoma. ADHD-type behaviours including impulsivity and ‘hypothalamic rages’. Autistic Style behaviours; cognitive delay/decline as the brain is constantly disrupted by epileptogenic signals. ‘As the child develops, typically between age 4 and 10, the epilepsy progresses to focal and generalised seizures. Short term memory and processing difficulties. Lack of sleep or disrupted sleep. Some patients present with endocrine issues including precocious puberty, other sex hormone disturbances, hypothyroidism, panhypopituitarism, growth deficiency hormone and hypothalamic obesity.

NEED Recognition that the (sometimes subtle) epileptic occurrences are seizureal and not behavioural or reflux for example. Patient requires an MRI scan to a specific protocol (thin slice) because some HHs are very small and subtle in their attachments and so are frequently missed by radiologists.

IDEALLY Where HH is suspected, an apparently normal MRI should be reviewed by a radiologist and neurologist with HH experience. Referral immediately upon diagnosis to a multi-disciplinary team at a specialist epilepsy centre.

Diagnosis

Often mis-diagnosed. Paediatricians do not recognize the laughter/crying episodes as seizureal. Eye and mouth movements may be dismissed as ‘tics’. Due to co-morbidities affecting behaviour and delays in cognition, patients are often initially diagnosed with ADHD, Autism or other learning disabilities and/or behavioural conditions. The significant percentage of HH patients who present with precocious puberty are often the patients that receive early diagnosis. EEG and VEEG unhelpful as gelastic seizures usually don’t show up.

NEED MRI scan to specific protocol. Experienced multi-disciplinary team able to address patient concerns /questions (frequent complaint: ‘my neurologist didn’t know much about it.’) Work-up to rule out endocrine issues; genetic testing to be considered as 5% of HHs are caused by Pallister-Hall Syndrome, a mutation in the Gli3 gene.

IDEALLY Specialist Panel review. Advice must be tailored to patient and type and attachment of HH, rather than offering the preferred surgical option of treating hospital. All options should be discussed to enable patient to make informed decisions.

Treatment

Precocious puberty is usually well-controlled through medication. Gelastic seizures are typically refractory to AEDs although secondary seizures may be better medically controlled. Surgery to disconnect or destroy the HH appears to be the only way of stopping or reducing the seizures permanently. Post-surgery, seizures and co-morbidities often require on-going management, and there may be side-effects from the surgery itself.

NEED Evidence suggests that the earlier the surgery, the better the outcome for the patient. In significantly cognitively impaired patients there is little point delaying to try different combinations of AEDs since these almost never have any effect on gelastic seizures.

IDEALLY Timely surgery. Pre-surgical protocols re endocrine, neuropsychological and ophthalmological function should be conducted to establish a baseline pre-surgery. Follow-up at regular intervals post-surgery

Surgery

Different surgical techniques aiming to disconnect or ablate the HH have all had some success, depending on the HH type. Gamma Knife surgery has a good success rate for smaller HHs, with low risk of complications. Laser ablation has had a good success rate also – more immediate cessation of seizures than GKS, but greater risk of complication. Stereotactic thermocoagulation has been effective in Japan and some European centres. Endoscopic or transcallosal surgery may be indicated for larger (giant) HHs.

NEED More than one surgery is often required. Even if surgery is successful in seizure control, patients likely to need long-term educational support – learning support assistant, occupational therapist, speech and language therapist – within a specialist environment. Surgery can cause significant side-effects, particularly endocrinological and/or memory issues which require long-term monitoring and managing. Puberty may trigger renewal/worsening of seizureal and behavioural symptoms.

IDEALLY Multi-disciplinary approach to continue, recognising that maximising quality of life means much more than seizure control. Team should support the child’s educators and should liaise with child and adolescent mental health services when necessary:– HH is a multi-faceted syndrome with many co-morbidities and should be managed as such. Careful transition from paediatric to adult services to support independence, social integration, employment and mental health:

Follow up

With a neurologist re the seizures but also there should be cognitive, endocrinological and neuropsychological follow up with annual/biannual testing. This is a multi-faceted condition which requires management and follow up from a team of specialists, particularly given the risk of surgical side-effects.