Characteristics of Hypothalamic Hamartoma (HH)

These characteristics of HH usually start in infancy or early childhood and its range, intensity and complexity can vary widely from individual cases.

Epilepsy; Hypothalamic Hamartomas present with isolated Gelastic seizures or a combination of Gelastic and other types of seizures.

- **Gelastic seizures** are characterised by episodes of laughter typically associated with a facial contraction that resemble a smile. Because the seizures resemble natural laughter, they might not recognised it as seizure initially thus delaying diagnosis. They usually manifest in infancy and are the first type to occur. The seizures may occur frequently including episodes during sleep. Adults and older children may have experienced the urge to laugh that can be suppressed or in milder form of epigastric discomfort. Dacrystic seizures can manifest, crying and facial contraction resembling a grimace and these seizures may occur simultaneously with Gelastic seizures. Gelastic seizure is a hallmark of epilepsy associated with HH.
- **Complex partial seizures;** the evolution usually takes place between the ages of 4 to 10 years. These seizures are features with impaired level of consciousness, focal motor convulsive and automatisms. It is not uncommon for features of Gelastic epilepsy to be retained in complex seizures. The manifestation of complex seizures will typically lead for the diagnosis of epilepsy in many children.
- **Generalised seizures;** Gelastic or complex seizures typically precede the onset of generalised epilepsy. Individuals may progress to general seizures, including tonic seizures, tonic-clonic and drop attacks.

- Despite the use of higher dose and several combinations of anti-epileptic drugs, seizure freedom or good seizure control is rarely achieved.
- Individuals who present in adulthood often have milder seizures that are not Gelastic
- Studies shows that the prevalence of epilepsy associated with HH is 1/200,000 however, the total percentage of sufferers who experienced seizures is unknown because some individuals present no symptoms or have mild presentations of HH.

Precocious Puberty

Symptoms often begin in early infancy. Precocious puberty is defined as pubertal onset before the age of 8 years in girls and 9 years in boys. Tuber cinereum hamartoma is the commonest cause of organic central precocious puberty. Central precocious puberty results from over secretion of gonadotropin releasing hormone (GnRH), which in turn results in over production of Luteinizing Hormone (LH) and Follicle Stimulating Hormone (FSH).

- Treatment; can be easily treated with medication. Consultation with an Endocrinologist (hormone doctor) experienced in precocious puberty is a must to discuss effective treatment and discussions of any possible side effects.

Cognitive and behavioural problems

Majority of patients with HH has some degree of cognitive impairment and significant proportion also has mood and behavioural issues. Cognitive impairment and behavioural problems are common in children with epilepsy associated with hypothalamic hamartoma (HH).

- Global developmental delay
- Speech and language
- Global memory deficits
- Emotional and behavioural deficits
- Attention deficit disorder
- Affective disorders
- Anxiety disorders
- Aggressive disorders/Conduct disorders

Although there are reports associating behaviour and cognitive problems to the frequency and severity of seizures from HH, there are still questions as to these problems are inherent or is a direct consequence of the seizure activity.