Hypothalamic hamartoma presenting with gelastic seizures, generalized convulsions, and ictal psychosis
Hypothalamic hamartomas (HH) are rare tumor-like heterotopic masses commonly found in the hypothalamus or tuber cinereum. They typically present in childhood with gelastic seizures and precocious puberty. Occasionally, the condition can be associated with intractable convulsive epilepsy and behavioral abnormalities. The most effective treatment is surgical removal of the HH. The aim of this case report is to alert the physician to early diagnosis of the condition and to prevent suffering in a manageable disease.

Case Report. This case is a 30-year-old right-handed Qatari female patient. The epileptic symptoms started at the age of 3½ years. Her preceding childhood development was unremarkable. In school, her performance was poor and she discontinued school in the second year after admission because of the seizures and cognitive difficulties. The clinical symptoms started with bouts of involuntary and irresistible laughter and dropping to the floor (gelastic seizures). Later in adulthood she started to develop other seizures of different semiology. Those seizures typically started with a brief period of oro-facial automatism in the form of repeated smiling followed by loss of consciousness and generalized tonic clonic convulsions. The ictus duration was usually less than 10 minutes and recurred up to 4 times per month. She had sustained many body injuries related to the seizures. Infrequently, she had minor seizures limited to an episode of repeated smiling for a few seconds or a drop to the ground. The seizures were not controlled by antiepileptic drugs despite taking twice daily doses of levetiracetam 1000 mg, carbamazepine 400 mg, and primidone (Mysoline) 250 mg. Occasionally, she developed sudden outbursts of aggressive and agitated behavior independently of the other seizures. She expressed rage, violence, and paranoid ideations. She destroyed furniture and TV apparatus in different violence incidences. The psychotic outburst might persist for up to one hour. After the attack had settled, she had no recall of how she had acted during the event. In-between the attacks, she depicted a quiet

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and a pleasant character. Erroneously she was thought
to have a primary psychotic disorder and was treated by
a psychiatrist for a long time, which had greatly delayed
the right diagnosis. Psychotropic agents were ineffective
in suppressing the aggressive psychotic symptoms. All
other forms of seizures were refractory to drug treatment.
Her neurological examination was unremarkable
except for a moderate cognitive impairment. Video-
EEG demonstrated non-localizing ictal pattern with
maximum amplitude in bifrontal regions. The 3T-MRI
demonstrated an oblong 1.2 x 2.5 mm, well demarcated
soft tissue mass, iso-intense to the grey matter, in the
region of the hypothalamus (Figure 1). The lesion was
partly intrahypothalamic with an extension below
the hypothalamus. No enhancement was seen on
the post-gadolinium images. An incidental small left
vestibular schwannoma was seen on the MRI (Figure
1). Based on the characteristic imaging features, lack of
enhancement and typical location, the diagnosis of HH
was considered. The tumor was surgically reached and
resected by a transcallosal endoscopic approach via a left
craniotomy (Figure 2). Histopathological examination
of the surgical material confirmed that the mass was a
hamartoma. She developed a transient diabetes insipidus
as a complication of the surgery. Postoperatively, she
was regularly seen at the outpatient epilepsy clinic for
neurological assessment and treatment. By the time
of writing this report, she was followed up for more
than one year. The seizure frequency had regressed by
more than 50% after surgery. The psychotic symptoms
had completely subsided. She continued to take
anticonvulsant drugs, and the dosages were adjusted as
needed.

**Discussion.** Le Marquand and Russell⁴ were
the first to describe HH in a child with precocious
puberty.⁴ They are rare developmental heterotopic
non-neoplastic tumor-like masses most commonly
found in the region of the hypothalamus and the
tuber cinereum. The HH are either intrahypothalamic (sessile), or parahypothalamic (pedunculated)
attached to the hypothalamus. Morphologically,
hamartomas are composed of glial cells and neurons
that morphologically resemble the normal cells of the
hypothalamus and tuber cinereum. The intrathalamic
hamartoma usually presents with epilepsy, while the
pedunculated variety presents with precocious puberty.¹
The location of the tumor, iso- or slight hypointensity
on T1-weighted images, hyperintensity on T2-weighted
images, and lack of gadolinium enhancement are
characteristic MR features that strongly support the
diagnosis of HH.⁵ Our patient presented with gelastic
seizures in her early childhood, which is the typical early
clinical presentation of sessile HH. Later in the course
of epilepsy, the gelastic component often becomes less
prominent and other types of generalized or partial
seizures may appear. The epileptic syndromes that may
develop in adolescence or adulthood include generalized
tonic clonic, secondary generalized tonic clonic, tonic,
atonic, and complex partial seizures.² The HH can lead
to impairment of cognition, which may interfere with
the learning process and schooling.⁶ Gelastic epilepsy in
HH is known to be associated with ictal and interictal
behavioral abnormalities mainly aggression, paranoid
ideation, and major affective disorders. Ictal psychosis
is usually episodic and associated with generalized
seizures.⁶-⁸ Interictal psychosis is more chronic however;
it has a benign course and better response to treatment.⁸
The seizures associated with HH are often described as
catastrophic because of its refractoriness to treatment
with anticonvulsants. However, the progressive
epilepsy, behavioral, and cognitive deterioration can be
ameliorated by surgical intervention. Several surgical
procedures are used to reach the HH.⁹ The tumor was
successfully removed in our patient by endoscopic

![Figure 1](image1.png)
*Figure 1 -* Patient MRI a) T2 weighted showing a hypothalamic hamartoma (arrow) and b) 3T showing an incidental small left vestibular schwannoma (asterisk).

![Figure 2](image2.png)
*Figure 2 -* An MRI of the brain after surgical resection of the hamartoma showing the left craniotomy (white arrow) and the transcallosal endoscopy tract (black arrows).
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resection, resulting in a remarkable improvement in epileptic seizures and psychotic symptoms after several decades of suffering.

In conclusion, the HH is a rare tumor that has characteristic clinical features and can cause intractable epilepsy of multiple semilogies. Unfortunately the diagnosis of HH, a condition that can be ameliorated by surgical intervention, is often delayed particularly in developing countries. A protracted course of suffering can be avoided if the condition is considered in every case of gelastic seizures or precocious puberty.

References


ILLUSTRATIONS, FIGURES, PHOTOGRAPHS

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