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Clinical Image

Open-Lip Schizencephaly in Adulthood

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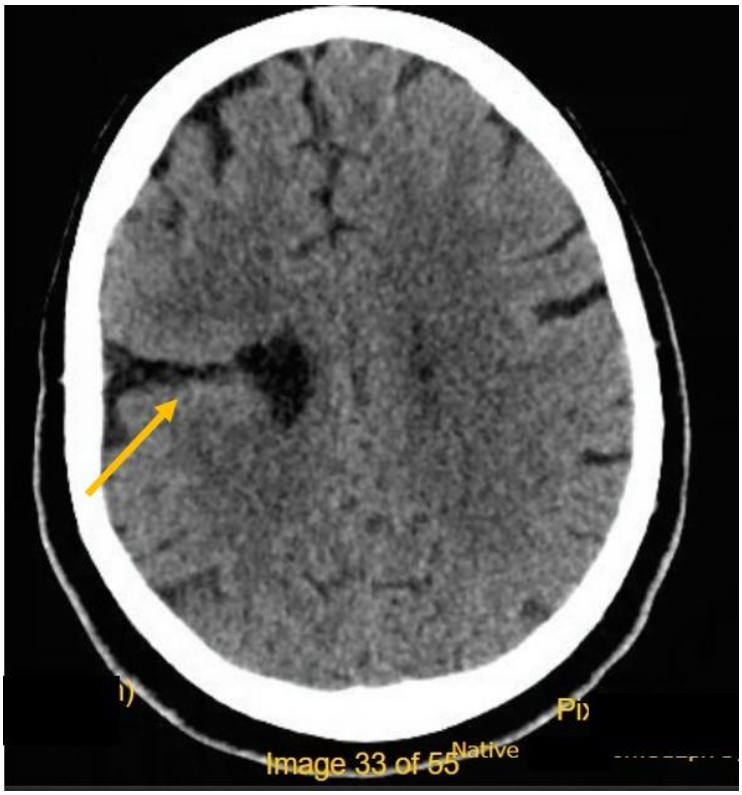

Figure 1: Axial cranial computed tomography scan showing a cleft filled with cerebrospinal fluid in the right cerebral hemisphere, consistent with open-lip schizencephaly.

A 47-year-old woman with left-sided hemiparesis of unknown cause since childhood, without other pathological history or medications. She had no contact with her parents, so familial and gestational history is unknown. She was referred to the emergency department after a seizure witnessed by relatives. Physical examination upon hospital admission revealed muscle strength graded 4/5 in the left hemibody, without other abnormalities. Laboratory tests were unremarkable. Cranial computed tomography revealed a pathway filled with cerebrospinal fluid surrounded by cortex, contacting the lateral surface of the right frontal lobe with the ipsilateral lateral ventricle, indicative of open-lip schizencephaly (Figure 1). The patient was treated with Levetiracetam 1000 mg divided

into two daily doses and referred to a Neurosurgery outpatient clinic for further evaluation.

Schizencephaly is a rare congenital defect in cortical development, with an incidence of 0.54 to 1.54 per 100,000 live births [1]. It is almost always sporadic, with only a few familial cases described. The condition is characterized by a cleft in the brain connecting the pia mater to the cerebral ventricles, with the cleft margins lined by gray matter. The causes are diverse, including teratogenic agents, prenatal infections, maternal trauma, or mutations in the EMX2 gene [1, 2]. Clefts may be unilateral or, more commonly, bilateral, and are classified into two types: type I, with closed lips (two overlapping cortices), and type II, with open lips (cleft filled with cerebrospinal fluid). Clinical presentation varies widely depending on the extent and location of the cleft, with commonly reported findings including muscle weakness, intellectual developmental delay, and epilepsy, which may only manifest in adulthood. Some patients are asymptomatic, and the malformation is discovered incidentally through imaging [3-5].

Diagnosis is imaging-based and can be achieved with computed tomography. However, due to its higher sensitivity and specificity, magnetic resonance imaging is currently considered the most suitable diagnostic tool [2]. Treatment depends on several factors, including the severity of signs and symptoms. It typically involves anticonvulsant therapy and physiotherapy. While most patients achieve good seizure control, some may develop refractory epilepsy, which carries risks such as sudden unexpected death in epilepsy (SUDEP). Cases complicated by hydrocephalus and elevated intracranial pressure may require surgical intervention [1].

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