Clinical Reasoning: An 11-year-old boy with language disorder and epilepsy

SECTION 1

An 11-year-old boy with recurrent seizure attacks and expressive language disorder was referred to our clinic. His initial attack was at the age of 2, when he had a sudden facial twitching, followed by a generalized tonic convulsion together with loss of consciousness, lasting a few minutes. The episode recurred every few months thereafter. In addition, he could never utter any verbal language, although he could understand what others said, as well as follow their commands.

Further questioning revealed that he was born after a full-term difficult delivery. The Apgar score was 5 at birth. He could walk at 14 months. He was in the third grade of elementary school at the initial visit, with a poor intellectual performance in contrast to his normal motor development. He was able to conduct most daily activities, including eating, dressing, bathing, etc.

On physical examination, although his hearing and comprehension were normal, he had no verbal response and could only follow commands by nonverbal means. He had some difficulties in chewing and swallowing solid food. He presented with an inexpressive face with a half-open and drooling mouth (figure 1). Moreover, he was unable to show his teeth, close his eyes, or protrude his tongue on command, yet he could smile at a joke, yawn, close eyes during sleep, and move the tongue unconsciously. There was no atrophy of tongue muscle or fasciculation. The pupillary, corneal, gag, and jaw reflexes were normal, and a right Babinski sign was elicited. His other motor and sensory systems were normal, as well as his coordination and gait.

The interictal EEG displayed scattered slow wave from posterior leads (figure 2). No epileptic discharge was recognized.

After the administration of 400 mg carbamazepine a day, he was free of seizures, but all the other symptoms remained.

Questions for consideration:
1. What is the differential diagnosis at this stage?
2. What is the probable topical diagnosis?
The interictal EEG displayed scattered δ wave with low to medium amplitude (A) and slow wave at 2 to 3 Hz (B) from posterior leads.
SECTION 2
The differential diagnosis at this stage might consist of the following common diseases:

1. Landau–Kleffner syndrome (LKS): since the combination of epileptic disorder and language disorder are chief complaints of the boy, the differential diagnosis might come to epilepsy-aphasia spectrum, especially LKS, which is an acquired verbal auditory agnosia combined with a subsequent disruption of expressive language due to prominent epileptiform activity. It usually occurs in children who are between the ages of 3 and 10 years with previous normal age-appropriate speech, and mostly a spontaneous remission in adolescence in parallel to the EEG improvement. Our boy has never developed speech ability and his auditory language comprehension is excellent, which do not support the diagnosis of LKS.

2. Congenital vocal organ diseases, such as ankyloglossia: these might contribute to loss of expressive language ability, but they cannot cover all the symptoms of the patient. Furthermore, the boy has undergone ear, nose, and throat examinations repeatedly without positive findings.

   The boy’s distinctive symptoms are persistent movement disorders of facial, lingual, pharyngeal, masticatory, and laryngeal muscles, with preserved brainstem reflexes, normal tongue muscle bulk, absent tongue fasciculation, and pathologic laughter. These suggest that the location of neurologic impairment is at the supranuclear level such as cortical or subcortical area, instead of the lower motor neurons, such as motor nuclei in brainstem, cranial nerve, neuromuscular junction, muscle, etc.

   There seems to be a paradox in the clinical symptomatology of the boy. He has lost the voluntary movement of orofacial muscles, while the reflexive or involuntary movement is unaffected, which makes the topical diagnosis more confusing.

Questions for consideration:
1. What should be done next to disclose the topical diagnosis?
2. Can the lesion detected be responsible for all the symptoms?
SECTION 3
Cranial MRI revealed abnormal signal intensity and significant symmetrical atrophy in bilateral temporal, parietal, and insular lobes. The abnormal signal intensity was mainly located in subcortical white matter, which was hypointense on T1-weighted imaging and hyperintense on T2-weighted imaging, consistent with leukomalacia and gliosis. The deeper sulcal portions showed more prominent volume loss than the superficial parts, making a mushroom-shaped lesion (figure 3).

With the assistance of MRI, topical diagnosis could easily be achieved. The symptoms of the boy should be attributed to bilateral opercular lesions. Operculum comprises the cerebral cortices from frontal, parietal, and temporal lobes overlying the insula. The motor nuclei of the 5th, 7th, 9th, and 10th cranial nerves in brainstem are innervated by bilateral corticonuclear tracts descending from motor cortices, and the 12th cranial nerve is controlled by the opposite corticobulbar tract. Therefore, a bilateral interruption between motor cortices and cranial motor nuclei of brainstem can lead to the suprabulbar palsy. In contrast, the spontaneous and reflexive controls are regulated by thalamus, hypothalamus, and extrapyramidal tract, which are not involved in this case confirmed by MRI, resulting in the normal reflexive movement.

Questions for consideration:
1. What is the probable final diagnosis?
2. What is the differential etiologic diagnosis?
3. Does epileptic attack have a role in the pathogenesis of the disease?
SECTION 4

Foix-Chavany-Marie syndrome (FCMS), first described in neurologic literature by Foix, Chavany, and Marie in 1926, is a rare cortical type of pseudobulbar palsy, usually resulting from damages in the anterior part of the operculum. Clinical manifestations mainly consist of facial diplegia, hypersalivation, dysarthria, and dysphagia. FCMS could link language disorder, orofacial movement disorder, and bilateral opercular lesions in the boy. The selective paralysis of voluntary fascio-flaccid movements is referred to as “autonomic-voluntary dissociation,” which is a distinctive feature of FCMS. The etiologic spectrum of FCMS includes, among others, CNS infections, congenital abnormality, epileptic disorder, stroke, vasculitis, head trauma, tumor, and neurodegenerative disease. Based on the supplemental medical history and MRI manifestation, CNS infections, head trauma, and stroke could be ruled out. The persistent clinical course, neither progressive nor reversible, is not in favor of the diagnosis of tumor, neurodegenerative disease, or vasculitis.

The differential diagnosis at this stage includes:

1. Perinatal hypoxic-ischemic encephalopathy−related FCMS: considering the history of asphyxia at delivery, along with the ulegyric pattern of MRI, we infer that all the symptoms might be secondary to perinatal hypoxic-ischemic encephalopathy, which might be a new etiology never covered in the previous literature.

2. Congenital FCMS: the patients with congenital type could have extra developmental abnormalities such as developmental delay, arthrogryposis, pectus excavatum, micrognathia, hearing loss, hemiparesis, or paraparesis. MRI and pathologic examination could reveal perisylvian polymicrogyria or schizencephaly, failure of opercular closure, periventricular gray matter heterotopias, or absence of the septum pellucidum. There is no obvious evidence in support of the congenital FCMS in our case.

3. Acquired epileptiform opercular syndrome: it was first proposed by Shafrir and Prensky. When strong, persistent epileptic discharge spreads to bilateral opercular cortices, interfering with the cortical function in perisylvian regions, seizure patients can develop FCMS symptoms, which will disappear with EEG improvement. However, the operculum syndrome in the present case is supposed to be irrelevant to epileptic discharge or seizure episode. The explanations are as follows: the paralysis of orofacial muscles precedes seizure; the symptoms are persistent, do not fluctuate, and are not relieved with seizure remission; and no epileptic discharge has been detected in the symptoms.

DISCUSSION

In the differential diagnosis of children with difficulties in speech output and epileptic attack, especially those with autonomic-voluntary dissociation, FCMS should be considered.

The clinical course, treatment, and prognosis of FCMS are diverse, depending on the heterogeneous underlying etiologies. In adults, FCMS is mostly induced by multiple successive strokes involving bilateral anterior operculum. In children, meningencephalitis, epilepsy, and congenital malformation are the most frequent etiologies.

Although it is usually called “anterior operculum syndrome,” the lesions of FCMS are not limited to operculum. Most patients have bilateral opercular lesions, while some show a single opercular lesion and a coexistent contralateral subcortical lesion, or bilateral subcortical lesions without opercular involvement. Some even result from the lesions verified by SPECT, which are invisible on MRI. Thus, if the opercular lesions are negative on MRI in those with suspected FCMS, the subcortical lesions involving corticobulbar tracts should be explored. And if there are no structural lesions on MRI, SPECT should be performed to identify the possible functional lesions.

On the basis of anatomical knowledge, it is generally believed that a unilateral lesion does not result in FCMS and a bilateral structural or functional impairment between motor cortices and brainstem cranial motor nuclei is necessary. However, Giraldo-Chica et al. have reported an FCMS case caused by unilateral right opercular ischemia affirmed by both brain MRI and SPECT, which proposes whether a unilateral structural and functional lesion is sufficient for FCMS. Further research is needed to investigate whether the contralateral lesion is invisible because of the limitation of technique, or because there is an anatomical variant that the corticonuclear tract is predominantly unilateral.

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Dr. Dong drafted and revised the manuscript, including medical writing. Dr. Zhou critically revised the manuscript for important intellectual content.

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