RARE CASE REPORT OF LATE IDENTIFICATION OF CORPUM CALLOSUM DYSGENESIS IN A CHILD WITH BILATERAL HEARING LOSS

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Abstract: *The corpus callosum is the most important relay connection between the two hemispheres. The malformation of the corpus callosum manifests as total agenesis or dysgenesis of various degrees and these afflictions have highly variable clinical expression. Current imaging investigative possibilities allow the early detection of any malformations even in the fetal period. The cases in which brain malformation, either solitary or within a syndrome, associates with sensory or neural hearing loss are very rare. The authors present the case of a child with severe bilateral hearing loss and dysgenesis of the corpus callosum. The retrocochlear feature of hearing loss and the late identification of the malformative pathology of the brain is what gives the case its particularities.*

INTRODUCTION

The corpus callosum (CC) is the largest structure of the white matter of the brain, gathering more than 190 million axons. From an anatomic point of view, from the anterior to the posterior, there are four parts in its structure: rostrum, genu, body and splenium. Its dimensions vary between genders and races, but there is evidence that women are more developed in this aspect. The transverse fiber tracts of the CC structure connect the two hemispheres and play a role in the integration of the motor, sensory and cognitive functions. The literature indicates the presence of distinct specializations of the two cerebral hemispheres but also an interhemispheric communication made possible by the key structure, represented by the corpus callosum.(1) The existence of this neural bridge allows the transmission of information to the hemisphere that was not stimulated. Due to this structure, the inhibitory function of a hemisphere over the other one develops and creates asymmetry between the cerebral hemispheres (i.e. for right-handed people, the left hemisphere is dominant, with an inhibitory function on the right hemisphere). The presence of this structure makes possible the lateralization of functions such as speech, for which the left hemisphere is responsible in most people. The structures involved in this process are the lower part of the frontal lobe and the posterior left temporal lobe.

The corpus callosum is involved in gnosis and praxic processes, but also in mechanisms of memory and consciousness. Recent studies have revealed a differential specialization of the two hemispheres in processing linguistic features (2), the right hemisphere is involved in speech processing and suprasegmental features of speech, while the left hemisphere in segmental analysis of syntax and lexical semantics.(3) Corpus callosum plays an important role in complex processes such as understanding and speech production, with a syntax and prosodic information exchange between the two hemispheres. The malformation of this structure is generally described in the literature as dysgenesis of the corpus callosum, including both total agenesis of corpus callosum (its absence) and partial agenesis with varying degrees of atrophy and even the absence of parts of the corpus callosum morphology. Regarding the incidence of the disease, in the general population it is below 1% and for population with cognitive disabilities it is around 2.3% (4), having a predilection for males.(5,6,7)

Agenesis of the corpus callosum (ACC) causes can be multiple: rubella infection, alcohol, cocaine, genetic factors such as those involved in trisomy 8, 13 and / or 18, Andermann syndrome, Aicardi syndrome.

ACC can be solitary or associated with other malformations of the brain, and from a clinical point of view, cognitive dysfunction may be present varying from mild to pervasive disorders. Epileptic seizures occur in approximately 50% of cases. Some subjects may have normal intelligence (8), but most do not.

Genetic counseling in such cases remains difficult given that radiological and genetic markers can not distinguish between symptomatic and asymptomatic subjects.

Prenatal diagnosis is routine ultrasound (week 20) and magnetic resonance imaging (MRI) scan (week 30).

Treatment is symptomatic, including psychotherapy, speech therapy and anticonvulsant treatment for epileptic seizures.

Questions that have no answers refer to the fact that although this isolated malformation of the brain, total or partial, can be detected prenatally by ultrasound or MRI, it is impossible to predict the degree of functional impairment. Future multicenter studies will probably find the answer to which genes are responsible for this abnormality of the brain.

CASE PRESENTATION

The authors present the case of a male child, aged 7 years, who has developed bilateral deafness since early childhood, for which bilateral hearing aid were fitted by age 3. The child was admitted to the audiology department for second opinion due to the absence of any progress in speech development and inconsistent use of hearing aids.

Clinically, it was noticed a child with a slight mental

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retardation, hyperkinetic, with an extremely poor vocabulary including words composed of repeating syllables or incomplete words. The child had reduced ability to understand speech and communication with other people was possible mostly gestual.

The audiology examination consisted in acoustic immittance, behavioral audiometry through the observational method and auditory brainstem evoked response under general anesthesia.

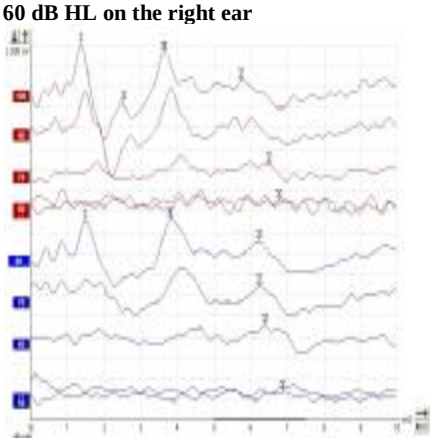
Tympanometry showed flat tympanograms (type B), which were consistent with serous effusion in both middle ears. Clinical examination of the ear showed bilateral retracted tympanic membrane with amputated light reflex. Due to the hyperkinetism of the child and his refusal to accept supraaural audiometric headphones, behavioral audiometry was possible in a free field arrangement with the loudspeaker located at 0 degrees azimuth. The minimum level responses ranged between 55 and 70 dB HL from 500 to 4000 Hz.

Electrophysiological assessment through brainstem auditory evoked response (BAER) was performed in the operating room under general anesthesia. To remove the conduction component of hearing loss, represented by serous effusion in the middle ears, a tympanostomy fluid drainage was carried out, followed by insertion of tympanostomy tube into the eardrum bilaterally.

The BAER test was performed with Eclipse 25 equipment (from Interacoustics), through air conduction, with ER3A intraaural transducers, collecting the evoked response in a two channel montage, the placing of the electrodes being FPz (high forehead), M1 (right mastoid) and M2 (left mastoid).

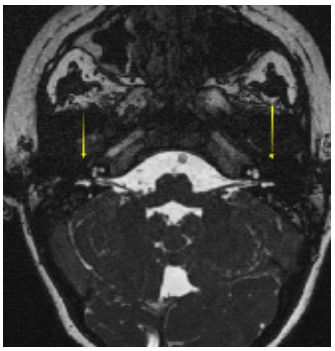
The stimulation parameters were short clicks in rarefaction polarity with a stimulation rate of 20.1 cicles/sec and bandpass filtering of the recording system from 150 to 3000 Hz. The results showed a moderate hearing loss with the objective threshold of wave V at 60 dB HL for the right ear and 50 dB HL for the left ear. The BAER recording analysis showed evidence of neural (retrocochlear) damage, by prolongation of I-V interval (4.77 msec compared to 4.4 msec maximum allowed) on the left side (figure no. 1).

Figure no. 1. click ABR recordings with prolonged I-V interval on the left ear (4.77 msec) and interaural I-V difference of 0.4 msec; I-V interval within norms on the right ear (4.37 msec); click tresholds at 50 dB HL on the left ear and 60 dB HL on the right ear



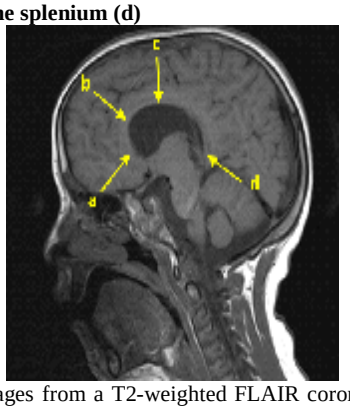
The interaural difference of I-V intervals was 0.4 msec. (0.3 msec maximum allowed). Given the suspicion of a left retrocochlear injury, the patient was referred for a brain magnetic resonance imaging (MRI) scan. The examination was performed on a Siemens Magnetom Avento MRI machine, with a magnetic field strength of 1.5 T and 0,8 mm slice thickness. It did not reveal the presence of any tumor mass in the left internal auditory canal or pontocerebelos angle, but showed severe dysgenesis of the corpus callosum, internal hydrocephalus with the dilatation of the lateral, third and fourth ventricles. The morphology of the cochleea and eight nerve was normal on both sides (figure no. 2).

Figure no. 2. T2-weighted axial flair acquisition shows normal content of internal auditory canal and cochleea on both sides



On the sagital T1 weighted view it was noticed corpus callosum abnormality with severe atrophy of the rostrum, genu, body and absence of the splenium (Figure 3).

Figure no. 3. T1 weighted mid sagital view shows severe atrophy of the rostrum (a), genu (b), body (c) and the absence of the splenium (d)



Images from a T2-weighted FLAIR coronal sequence showed enlarged lateral ventricles, third ventricle (Figure 4) and fourth ventricle (figure no. 5).

Figure no. 4. T2-weighted Coronal Flair slice showing hypertrophy of the lateral ventricles (a) and third ventricle (b)

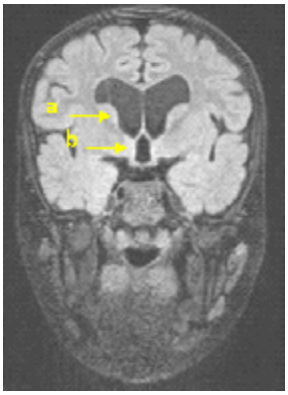
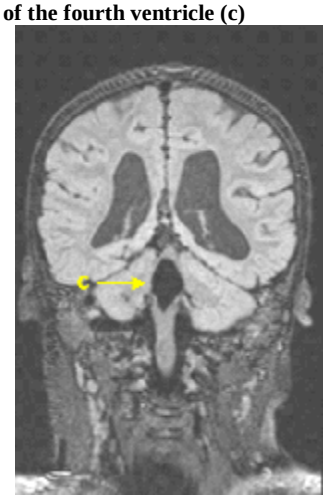


Figure no. 5. T2-weighted Coronal Flair slice showing hypertrophy of the fourth ventricle (c)



DISCUSSIONS

The authors reported an interesting case of severe dysgenesis of the corpus callosum with behavioral disorders and cognitive deficit associated with bilateral hearing involving permanent damage of the cochlea in both sides but also neural (retrocochlear) dysfunction in one ear.

This pattern of retrocochlear damage can be attributed to the compresive effect of the cerebrospinal fluid within the pontocerebellar cistern on the auditory nerve, but it can not be ruled out an intrinsec characteristic of the hearing loss associated to the malformation of the most important structure of the white matter in the brain.

Unfortunately, the malformation was not evidenced in utero, and the late diagnosis was detrimental to the possibility of applying specific recovery therapies until the age of 6 when the cerebral plasticity starts to decline significantly. Regarding the benefit of hearing aids, it is extremely limited, on the one hand due to the characteristic of neural deafness on the left ear, on the other hand because of the absence of communication between brain hemispheres that limits speech understanding but also its production. Other consequences of central auditory processing deficit are related to the difficulties that the child has on spatial localization tasks and dichotic listening.

These issues must be explained to the family in a manner that makes their expectations of hearing aids benefit and child's recovery be more realistic. Provision of conventional hearing aids may increase the child awareness to the sounds but the associated behavioral disorders disrupt a systematic wearing of the devices.

Concerning the prevalence of hearing loss in newborns with corpus callosum agenesis, there are no conclusive data reported in the literature so far. Some authors published isolated cases of association between agenesis of the corpus callosum and Mondini dysplasia (9) or congenital hearing loss in combination with partial agenesis of corpus callosum with hydrocephalus and arachnoid cyst.(10) For the central processing of auditory information that refers to specific tasks like attention, detection and understanding of sounds it is required integrity of structures of the brainstem to the cortex. Audiological monitoring of central auditory pathways is recommended in all cases of children with dysgenesis of corpus callosum even if they have a normal function of inner ears detected by otoacoustic emissions screening technique at birth.

CONCLUSIONS

Although this malformation may be solitary, related to maternal consumption of alcohol and cocaine mostly, or syndromic appearance in 25-32% of patients (11), there are very few cases reported in literature with clinical manifestation of bilateral sensorineural hearing loss. This is a very rare case reporting the association of severe corpus callosum dysgenesis, internal hydrocephalus and bilateral hearing loss involving malfunction of both cochlea and brainstem audithory pathways. The pattern of inheritance is probably autosomal recessive.

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