Georgian Journal of Neurosciences, 1-3 (2004) 41-45

AGENESIS OF CORPUS CALLOSUM: A CASE REPORT

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Accepted 16 February, 2004

Abtsract

Patient with complete callosal agenesis, male, aged 7, displayed cerebral disconnection symptom as deficit in verbal identification of objects, palpated by the left hand. Compensatory mechanisms do not substitute efficiently for callosal pathway in connecting lefthemispheric language centers with somato-sensory regions of the right hemisphere. It is not clear, whether insomnia in this patient could be ascribed to the absence of callosal system, however, clinical, EEG and MRI examination did not reveal other visible reasons for sleep-wakefulness disorders.

Key words: corpus callosum, agenesis, hemispheres, callosal system, insomnia

Introduction

Agenesis of corpus callosum by its origin is related to the disorders of the formation of structures, deriving from the anterior telencephalic wall.

During the 5th week of embryo development the telencephalic vesicles (future hemispheres) arise as lateral evaginations of the prosencephalic cavity. Hemispheres are formed as a result of the backward growth of the lateral vesicles by the 7th – 8th weeks. Forebrain commissures and related structures originate from *lamina terminalis*, which develops from anterior telencephalic wall (originally arising from the wall of prosencephalic cavity) and extends from the optic chiasm up to the velum transversum. Its dorsal part is known as the *lamina reuniens* of Hiss.

The dorsal part of the *lamina reuniens* forms so-called *sulcus medianus telencephali medii* by infolding between the growing vesicles. *Massa commissuralis* – the fused tissue of the *sulcus medianus*, gives rise to corpus callosum, hippocampal commissure and fornix as well as to the hippocampus and archicortex. Callosal fibres appear at about the third month of embryo development and attain final configuration two months later. Ventral part of *lamina reuniens*, adjacent to *lamina terminalis* gives rise to anterior commissure, septal nucleus and rhinencephalon [5, 15, 21].

Thus, it is obvious that early injury to the anterior telencephalic wall may lead to commissural malformation, as well as to underdevelopment of olfactory structures and to holoprosencephaly. The localization and extent of injury determines which structures will be involved in the pathology. Agenesis of corpus callosum, anterior commissure and the hippocampal commissure may stem from the defect in the entire *lamina reuniens*, whereas in the case of a more dorsal injury, anterior commissure could be spared. Focal damage to the posterior midline at a later stage of morphogenesis may cause partial callosal agenesis, while in cases of more ventral injury corpus callosum is spared [5, 17, 24]. Corpus callosum is always absent in holo-

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prosencephaly [5]. Fibrous membrane replaces corpus callosum in agenesis [18] and the dilated third ventricle may open directly onto the surface of the brain. Misdirected callosal fibres (less than those in normal corpus callosum) could be found originating in the frontal lobe and terminating in the occipital lobe of the same hemisphere [5]. Associated abnormalities commonly found with the loss of corpus callosum are as follows: radial arrangement of supracallosal gyri, polymicrogyria, absence of the cingulate gyrus, the septum pellucidum and the hippocampal commissure.

Callosal agenesis is reported in children with Joubert syndrome [27] while callosal agenesis in combination with multiple cysts, subtle eye abnormalities and skin defects is considered a syndrome, caused by deletion in the chromosome 14.q11.2 region [28].

At the same time chromosomal anomalies, representing trisomy of chromosome 8, are believed a cause of corpus callosum agenesis or hypoplasia, concomitant to craniofacial anomalies [7, 8].

It was established earlier that callosal agenesis may occur as an isolated phenomenon [14, 20].

It is believed that input provided by callosal axons is necessary for a normal development of the neocortex [1]. Some spatial and perceptual-motor problems, as well as difficulties in nonverbal reasoning have been reported in young female patients with callosal agenesis [23]. Bilateral motor discoordination due to the callosal agenesis was shown in adults [15] and in children as well. Epileptic disorders are commonly observed in patients with callosal agenesis [3, 4, 7, 11, 19]. Hence, seizures may be responsible for mental retardation at least in some cases of callosal agenesis. In fact, it is an old and still unsolved problem whether the agenesis of the callosal fibres by itself may cause determined psychic abnormalities. Comparative study of patients with isolated and non-isolated callosal agenesis is of special import in this respect.

Out of three patients of Gon-Alves Ferreira and colleagues [12] with isolated corpus callosum agenesis one is reported asymptomatic, another displayed slight language delay and the third had a mild developmental delay, whereas five subjects with non-isolated callosal agenesis suffered from psychomotor developmental delay, epilepsy, hemiparesis, ocular apraxia and macrocephaly.

Chacko and colleagues [4] reported 22 cases of callosal agenesis out of 2164 children who underwent CT examination. Most cases were not syndromic. Abubacker and colleagues observed the adult with interhemispheric cyst and partial corpus callosum agenesis without any clinical symptoms related to the entity [2].

"Human Cerebral Disconection Syndrome" rising after surgical section of the corpus callosum includes specific symptoms ascribed to the defiiciency of interhemispheric exchange of information via callosal system. Hemispheres work as a separate units and visual, auditory, or somesthetic stimuli, confined to one hemisphere, do not reach appropriate centers of the opposite halfbrain. As a result, acallosal patients fail in intermanual transfer task performance, they display apraxia in a left hand in response to verbal command perceived by the right hemisphere. Patients with callosal section fail to name objects, perceived by the right hemisphere [10]. It is still unclear, however, whether patients with callosal agenesis display the symptoms, as callosotomized patients do.

To extend data on cerebral disconnection syndrome in callosal agenesis,

present report is focused on neuropsychological examination of the patient with complete agenesis of corpus callosum.

Material and Methods

Patient, male, aged 7, underwent MRI (*conducted at the Institute of Radiation* and Interventional Diagnostics, Tbilisi) investigation and was diagnosed with isolated complete agenesis of corpus callosum.

Anamnesis vitae: on seventh month of pregnancy mother had an infectious disease (grippe), presumably causing premature birthgiving. Infant experienced two attacks of febrile seizures at a first year of life and has been treated with sedatives till the age of 2. Child started walking independently at an age of 14 months and speaking fluently at an age of 2. Child could not sleep last 4 years. Parents mention neither emotional nor behavioral disorders and addressed physician exclusively because of insomnia.

At the same time according to mother: "Sometimes he lapses into a doze and does not respond when I call him, however, this lasts a couple of seconds, not more". The EEG examination (conducted at the Institute of Physiology, Tbilisi) did not reveal epileptic activity of brain. Mild enlargement of reflexogenic area, as well as moderate reinforcement of basic reflexes have been observed by neurologist. Vision was found normal.

To reveal callosal defficiency clinical examination was followed by a series of special tests designed to study interhemispheric transfer and integration of somesthetic and motor functions. The following tests were employed: transfer of kinesthetic information, bilateral coordination, intramanual and intermanual localization of touch, intermanual matching of objects and stereognosis.

Results and Discussion

Subject was found perfectly able to cross-replicate hand postures in both, right-to-left, as well as left-to-right directions. Bimanual coordination as well as touch localization and intermanual matching were performed without errors. The results of stereognosis task performance are summarized in Table 1. All objects were correctly named when placed in the right hand of the subject. Total 3 objects out of 8 presented were correctly named when placed in the left hand of the subject. When two objects were presented simultaneously to the two hands, successive verbal report revealed preference of the right hand in all 4 trials applied to the subject.

Unlike some previously described cases [9, 11, 16, 23, 25, 26] agenesis of the corpus callosum in our patient did not affect intermanual task performance, as well as bimanual coordination.

The compensatory brain mechanisms suggested in this context are increased use of either ipsilateral or subcortical

Table 1

Errors in the stereognosis test

Hand	*Objects*							
	pencil	*ball*	*glasses*	*eraser*	*matchbox*	*spoon*	*fork*	*key*
Left	–	+	+	–	–	–	+	–
Right	+	+	+	+	+	+	+	+

" + " – correct response, "– " – error.

pathways as well as participation of other neocortical commissures, especially the anterior commissure [10, 13]. At the same time, our findings do not confirm the suggestion that agenesis of the corpus callosum is asymptomatic [2, 24].

Results obtained in present study point at partial disconnection between two hemispheres. The results of stereognosis task performance attract special attention in this respect. Evidently, callosal pathway is crucial for connecting language centers of the left hemisphere with somato-sensory regions of the right half-brain. Every clinical case of callosal agenesis has its individual symptoms and usually we reveal rather differences than the common signs when comparatively analizing the cases of callosal agenesis.

In sum, patient with complete callosal agenesis, male, aged 7, displayed cerebral disconnection symptom as deficit in verbal identification of objects, palpated by the left hand. Compensatory mechanisms do not substitute efficiently for callosal pathway in connecting left hemispheric language centers with somato-sensory regions of the right hemisphere. It is not clear, whether insomnia in this patient could be ascribed to the absence of callosal system; however, clinical, EEG, and MRI examination did not reveal other visible reasons for sleep-wakefulness disorders.

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