Clinical, Radiological and Laboratory Features in Subjects with Complete Agenesis of the Corpus Callosum

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Abstract

Introduction:

The corpus callosum is a connecting structure between the two cerebral hemispheres. Its development occurs between 8th and 20th week of gestation. Anomalies of the corpus callosum are divided into malformative or acquired anomalies and may be isolated or associated with other cerebral malformations. It is commonly admitted that the presenting signs or symptoms in individuals with corpus callosum agenesis are due to concurrent brain abnormalities and that isolated is essentially asymptomatic. The corpus callosum agenesis is a common component in some malformative syndromes, frequent in another, and occasional in many of them. The corpus callosum agenesis has been reported in many chromosomal aberrations and less frequently in inborn errors of metabolism and neurocutaneous diseases. The diagnosis of an alteration of the corpus callosum is fundamentally based on neuroradiological examinations.

Methods:

In our study we evaluate the major clinical, radiological and laboratory findings, the psychomotor development, and the electroencephalographic and neuroradiological features in a group of 14 children with complete callosal agenesis, trying to single out how the associated cerebral malformations interfered with the clinical manifestations. It was not possible to highlight a distinctive symptom indicative of the presence of a corpus callosum anomaly;

Results: nevertheless in 7/14 children there were a psychomotor delay and mental retardation of varying degree. 2/14 of our subjects presented epilepsy, and EEG showed generalized high voltage_ slow wave and generalized spike wave in 3 patients. All patients had normal thyroid function test, routine laboratory test and urine and blood amino acid level. Children affected by isolated callosum anomalies did not present any significant clinical manifestations.

Discussion:

Finally, we conclude that the determinating factors in the clinical-electroencephalographic picture as a whole and especially in different presentations of our patients are associated to the callosum anomalies.

References

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