A CASE OF SCHIZENCEPHALY WITH POLYMICROGYRIA

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A case of extensive bilateral frontotemporal schizencephaly is alleged – more extensively in the left hemisphere – which associated with polymicrogyria. The cortical anomaly was discovered only incidentally by MR examination in a 22 year-old man who suffered from headache due to a mild head trauma. Neurological examination proved to be negative. He had no complaints or symptoms a few weeks later. The developmental anomalies in corticalisation are shortly overviewed in this group together with the possible causing factors. It has been emphasized the importance of the precise intrauterine and/or postpartum differential diagnosis between schizencephaly, porencephaly and other failure in corticalisation.

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SCHIZENCEPHALIA ÉS POLYMICROGYRIA ESETE

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The development and discoveries considering the molecular biologic bases of malformations in the corticalisation have rendered a new classification of the cortical malformations\textsuperscript{1, 2}. Particularly the use of various neuroimaging methods in every day practice has raised the need of the distinction between diffuse and focal/multifocal malformations porencephaly and schizencephaly respectively\textsuperscript{3}. The precise diagnosis of cortical malformations based on the stage of development (cell proliferation, neuronal migration, cortical organization) will enable practicing physicians, neurologists and neuroradiologists to obtain a better conceptual understanding of the clinical characteristics and to make a more accurate prognosis without the need for brain biopsy\textsuperscript{4, 5}.

Our case is a very useful example for the demonstration of an incidentally discovered developmental cortical anomaly which appears worthwhile communicating.

History

Gy. B., a 22 year-old man appeared for neurological examination due to a headache. This complaint had been caused by a head trauma suffered two days earlier during a game of football. He felt dizzy for several hours. His birth and development were normal. He had successfully completed industrial school and then worked abroad for two years without any complaints. The examinations did not reveal any physical (blood pressure, pulse) and neurological signs. Fundoscopy and the routine electroencephalography displayed no pathological change. His psychic and mental state appeared to be normal, as did his memory.

Neuroimaging (MR) disclosed a significant cortical developmental abnormality characterized by an extensive defective corticalisation. The defects included the almost complete “absence” of the left frontal and temporal lobes apart from the posterior halves of the superior and part of the middle frontal gyri, precentral gyrus, the anterior two-thirds of the temporal lobe and the inferior quarter of the parietal lobe. Similar changes had occured in the right hemisphere but the area involved was significantly smaller in extension: Polar and basal sections of the

Figure 1. MR examination (T\textsubscript{1} sequentia) demonstrates the lack of parenchyma on six frontal sections

Figure 2. The MR examination (T\textsubscript{2} sequentia) show the extension of the same lesions in bilateral horizontal sections
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The deficiency of parenchyma was filled by cerebrospinal fluid. The cortex of the parts affected appeared to be continuous characterized by polymicrogyria which showed a moderately smaller diameter compared to the normally developed regions it was inferred that the structure of the microgyria may be four- or six-layered (Figures 4.). The volume of the white matter near the damaged cortical region also decreased, and the basal ganglia, internal capsules on the left side became smaller to such a degree that the latter were indistinguishable except the head of the caudate nuclei. The anomaly extended on the left almost to the middle part of the Sylvian fissure respectively to the level of the anterior third of the thalamus and ended in the right hemisphere more forward as depicted on the horizontal and frontal sketches (Figure 5.). The subcortical structures disclosed more moderate changes on the right side so they were more distinctly visible. The midline structures such as the optic nerves, callosal body, fornix, third ventricle, infundibular areas etc. were in their normal position. The ventricles were medium in size and normal in form, but the left lateral ventricle was slightly narrower than the right. The brain stem was normal. The size of the right cerebellar hemisphere was smaller in size than the left but the folia did not show atrophy.

Neurosurgical examination found no cause for surgical intervention due to the supposed disturbance in the cerebrospinal fluid passage. They were in the opinion that the headache was caused by a mild – and probably transitory – increase in the cerebrospinal fluid pressure.

The final clinical diagnosis was schizencephaly polymicrogyria (mainly bilateral frontotemporal localisation) with no clinical symptoms.

The complaints disappeared after three weeks of medical treatment. Genetical testing of the patient and his parents was planned but their consent was not obtained, so a chromosomal variant of this anomaly may not be established. The parents even refused a routine MR examination. The last examination of patient revealed no complaints or symptoms after two years.

Discussion

Malformations in cortical development may be divided into three groups: abnormalities resulting from the disturbances of cell proliferation, neuronal migration and cortical organization. Each of these three major abnormality types may be further clas-
sified into the diffuse and focal or multifocal malformations (Table 1). The molecular biology cleared more genetic variants based on different mutations of the same causative genes resulting differences in phenotype such as the gene affecting the protein function, different dosage of the same mutation in the same gene and the different effects of the same mutation and dosage caused by unknown factors1, 2.

In some cases of polymicrogyria (six patients), large deletions in the DiGeorge region of chromosome 22q11.2 had occurred and two parents of patient showed the same deletion, but the features of the polymicrogyria were different. These findings support the role of some modifying factors which may cause variants in phenotype. Mosaic mutations such as these, therefore, have a mild phenotype with low inheritance risk which may be of great importance in genetic counseling. Some authors have found the occurrence of a mutation in the EMX2 gene to be a causing factor in some cases of schizencephaly, but this finding has not been confirmed1, 7.

According to the revised classification of the malformations the polymicrogyria and schizencephaly together may be classified into the third group caused by abnormal cortical organization including later neuronal migration under the point A. 2. a. (Table 1)4, 5. Harding and Copp1 have sug-
1. References

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