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SHORT REPORT

Hemimegalencephaly and normal intellectual development

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Abstract

Hemimegalencephaly is a rare congenital malformation characterised by overgrowth of one hemisphere. Although it is commonly thought to be associated with neurological deficits, developmental delay, and intractable epilepsy, the clinical expression of hemimegalencephaly, can vary widely. This patient was neurologically and neuropsychologically normal apart from rare partial seizures.

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Hemimegalencephaly is a congenital cerebral abnormality consisting of unilateral hypertrophy of the brain. Most patients with hemimegalencephaly present with contralateral hemiparesis, mental retardation, and refractory partial epilepsy. Unlike most reported cases, our patient was otherwise neurologically normal.

Case report

The personal and neurological history of this 19 year old patient was normal; pregnancy and birth were uncomplicated. He had a normal psychomotor development and no learning disability. His parents (who were unrelated) and sister and brother were in good health. When he was 14 years 5 months old, brief episodes of sudden repetitive jerks developed in the left arm; these attacks occurred mostly after awakening and recurred daily. A month later he was admitted to hospital because one such episode was followed by a tonic-clonic seizure. A CT scan obtained on admission (fig A) demonstrated a pathological right hemisphere, which appeared increased in size, with high lucency of the white matter. This was interpreted as oedema of the right lobe and despite his normal neurological condition the patient was treated with high doses of mannitol and corticosteroids. He was discharged on a regimen of phenobarbitone.

Three months later he was referred to us for further investigation. On admission, his neurological status was normal. Neither somatic hemihypertrophy nor skin lesions were evident. Biochemical findings were within the normal range. Analyses of CSF were normal. Sensory conduction in the left ulnar and sural nerves, visual evoked potentials and somatosensory evoked potentials to right limb stimulation were all normal, but the evoked potentials to left limb stimulation were abnormal. Cortical response to stimulation of the left common

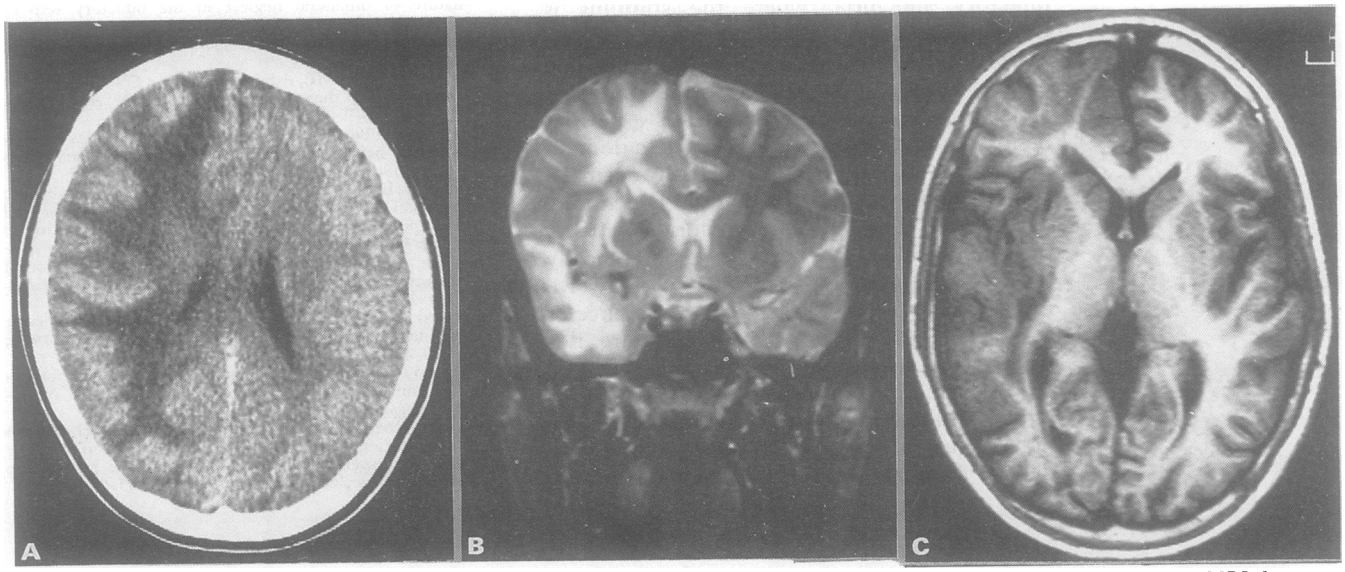

Figure A) Axial CT scan shows an enlarged right hemisphere. Note the diffusely hypodense right white matter. B) Coronal SE 1994/100 MRI shows a moderate enlargement of right hemisphere, increase in white matter, with prolonged T_2 relaxation time; at this frontal level cortical gyri and sulci appear normal. C) Axial IR 1615/30 MRI through basal ganglia shows abnormal gyral pattern in posterior temporal region. Note that in B) and C) the straightening of right frontal horn of lateral ventricle which is characteristic of hemimegalencephaly. (In all figures the right side of the brain is on the left of the figure).

peroneal nerve was absent and response to stimulation of the left median nerve was delayed and decreased in amplitude. Transcortical magnetic stimulation showed normal pyramidal conduction on both sides. EEGs showed fast beta rhythms in the right frontal and central regions, with frequent spike and spike-and-wave complexes in wakefulness and sleep. Seizures recorded on video and EEG were characterised clinically by brief, arrhythmic muscle jerks in the left arm, accompanied electrically by right frontal spike-and-wave complexes that became bilateral.

The results of neuropsychological tests were normal; the patient had an IQ of 115. MRI (figs B and C) demonstrated overgrowth of the right hemisphere, which was 1·2 times larger than the left. The right hemispheric white matter was increased and had a prolonged T2 relaxation time. The cortical gyri and sulci in the frontal, anterior temporal, and occipital regions appeared normal. In the posterior temporal and parietal regions the grey matter was slightly increased in size, with multiple macrogyri. The grey-white matter junction was distinct in this area only.

Although phenobarbitone and carbamazepine failed to control the seizures, they disappeared with valproate and have not recurred. The patient's current neurological examination confirms the absence of abnormalities. He is now working as an office machine technician. Recent EEGs show less paroxysmal activity: this now occurs only during hyperventilation and sleep. The fast beta rhythm is still present in the central and parietal regions. A second battery of neuropsychological tests, given four years after the first, showed a verbal IQ of 113 and a performance IQ of 114; visuospatial and superior cognitive functions were normal.

Discussion

Hemimegalencephaly is a dysplastic lesion characterised by an overgrowth of one hemisphere. The cerebral cortex of the megalencephalic hemisphere is generally abnormal and commonly has large pachygyric areas, at times associated with polymicrogyria. The white matter is also abnormal. The neuroimaging features of hemimegalencephaly have been adequately described.[1-3] Mental retardation and contralateral hemiparesis are generally associated with early epilepsy that is highly resistant and at times mandates surgery; the treatment usually chosen is hemispherectomy.[4,5] Nevertheless patients with slight mental retardation and non-refractory epilepsy with minimal neurological signs and without epilepsy have been observed.[6] The clinical expression of hemimegalencephaly thus varies widely. A similarly wide variation also appears likely in the anatomical lesions.[7]

In describing 12 patients with hemimegalencephaly, Barkovich and Chuang[3] distinguish two anatomical patterns depending on white matter amount and morphology. In all the cases described, the cerebral cortex was markedly abnormal with diffuse areas of pachymicrogyria or polymicrogyria. Barkovich and Chuang did not attempt to correlate the anatomical appearances of their patients with the clinical features. Our patient had overgrowth of one hemisphere, in which the white matter was increased and had prolonged T2 relaxation time on MRI. Unlike Barkovich's patients, the cerebral cortex conformed to an altogether different pattern: the frontal, anterior temporal and parietal lobes had normal gyri and sulci. The marked abnormalities affected only a small area of the posterior, temporal and parietal lobes. In our opinion, the preservation of normal cortex largely explains the patient's normal neuropsychological status. We observed that the neuromental development in hemimegalencephaly is notably worse in patients with more pronounced agyria or pachygyria[7] and probably correlates with the cortical pattern of malformation. Routine neurophysiological studies, including magnetic transcranial stimulation and visual evoked potentials, confirmed the integrity of the central and occipital regions. Abnormal right somatosensory evoked potentials to contralateral limbs corroborated anomalies in the parietal cortex. The normal neuropsychological testing results also demonstrated the integrity of higher symbolic functions. It is unclear why the abnormal MRI white matter signal was not expressed clinically.

Stable left lateralisation (contralateral to the hemimegalencephaly) and arm localisaton characterised our patient's seizures. They manifested with single arrhythmic jerks of variable frequency and amplitude involving different muscle groups. This seizure pattern differs from the partial motor seizures of rolandic origin, which are rhythmic, localised in one muscle group alone or successively involve more groups according to the somatotopic distribution of the cerebral cortex. It is, however, typical of focal cortical myoclonus, earlier reported in association with contralateral focal pericentral lesions (tumour or cicatrix) or contralateral atrophy, as seen in infantile hemiplegia.[8] More recently, Kuzniecky *et al*,[9] have described focal cortical myoclonus in three patients, who had focal dysplastic lesions, such as central macrogyria.

Apart from the epileptiform abnormalities, the EEGs of our patient were characterised by right central and parietal beta activity of low voltage. Patients with neuronal migration anomalies typically exhibit fast background rhythms[10] consisting of continuous alpha and beta activity. In bilateral malformations such as lissencephaly the activity is widely distributed over the two hemispheres; in focal dysplastic lesions it is lateralised or localised.

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