Neuroradiology (2001) 43: 792–793

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MRI in nonketotic hyperglycinaemia: case report

Received: 5 July 2000
Accepted: 27 September 2000

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Abstract We present a girl with proven nonketotic hyperglycinaemia. The pathological findings on MRI were brain atrophy with thinning of the corpus callosum and delayed myelination of the cerebral hemispheres, particularly the parietal lobes.

Keywords Nonketotic hyperglycinaemia · Magnetic resonance imaging

Introduction

Nonketotic hyperglycinaemia (NKH) belongs to a group of disorders of amino acid metabolism and is inherited as an autosomal recessive trait. The inborn defect in the glycine cleavage system leads to elevated levels of glycine in brain and cerebrospinal fluid which causes neurological impairment and myelin vacuolation. Imaging data concerning this condition are scanty. We present cranial MRI of a 14-month-old girl with proven neonatal-type NKH.

Case report

At the age of 3 days a girl developed seizures and apnoea; meningitis was diagnosed and treated in the intensive care unit. In the second month of life nonketotic hyperglycinaemia was diagnosed. At the age of 14 months the patient presented with seizures and psychomotor retardation.

MRI was performed at 0.5 tesla, with 7.5 and 5 mm spin-echo T1-, proton density and T2-weighted images in axial, coronal and sagittal planes. Advanced cerebral and cerebellar atrophy was found. Enlargement of the ventricular system was more pronounced than that of the subarachnoid spaces, especially in the cerebrum (Fig. 1 a, b); the corpus callosum was markedly thinned (Fig. 2). The infratentorial structures were normally myelinated, as were the internal capsules. Delayed myelination was observed in the cerebral hemispheres with areas of un- or possibly demyelinated white matter in the parietal lobes (Fig. 1).

Discussion

We found few reports concerning MRI in nonketotic hyperglycinaemia [1, 2, 3]. Atrophy and delayed myelination [1, 3] and thinning of the corpus callosum [1, 2, 3]

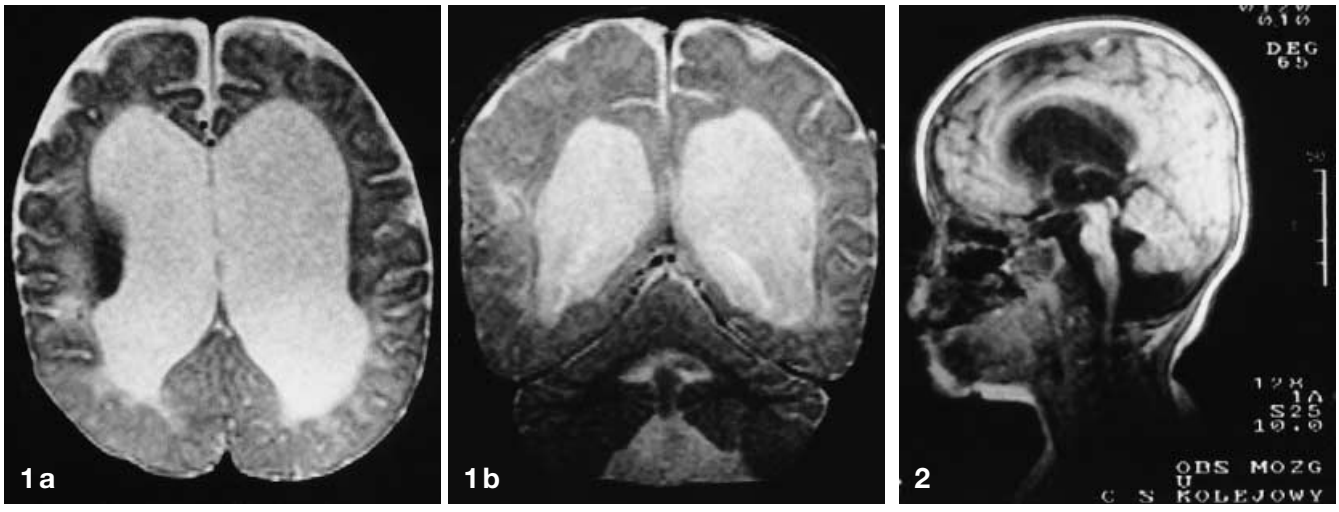

Fig. 1a, b T2-weighted images **a** axial, **b** coronal: there is marked dilatation of the supratentorial ventricular system, with moderate cortical atrophy, delayed myelination of the cerebral white matter and foci of un- or demyelinated white matter in the parietal lobes

Fig. 2 A T1-weighted image shows marked atrophy of the corpus callosum

were reported. Myelination of the cerebellum and brain stem was normal [1]. The MRI findings in our case are consistent with these data.

Acknowledgements This work was supported in part by grant 4P05B01311 from the State Committee for Scientific Research of the Republic of Poland to M. B. F.

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