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Brain MRI features in late-onset nonketotic hyperglycinemia

Achados de RM de crânio na hiperglicinemia não-cetótica de início tardio

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A 22-year-old woman presented with learning disability and gait unsteadiness since adolescence. Medical history revealed episodes of encephalopathy and myoclonic jerks associated with intercurrent infections. Examination showed spastic paraparesis, ataxia and optic atrophy. Neuroimaging revealed agenesis of corpus callosum (Figure). Plasma amino acid analysis disclosed elevated glycine levels with an increased cerebrospinal fluid:plasma glycine ratio.

Nonketotic hyperglycinemia (MIM #605899) is an autosomal recessive disorder of glycine metabolism caused by a defect in the glycine cleavage system with three different clinical forms: neonatal, infantile and late-onset with heterogeneous brain malformations, such as abnormal corpus callosum, gyral malformations and enlarged ventricles[1,2].

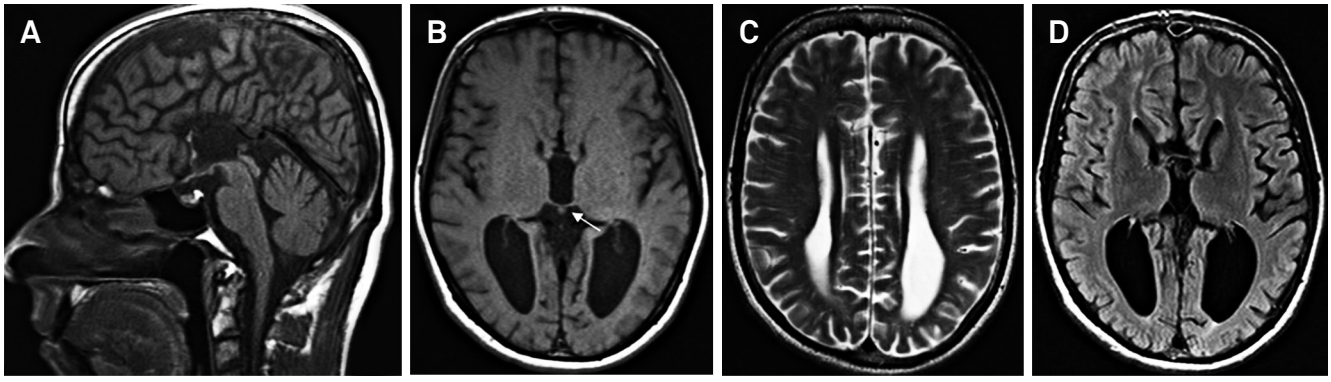

Figure. Neuroimaging findings in nonketotic hyperglycinemia. Sagittal and axial brain MR images disclosing nearly complete corpus callosum agenesis (sparing only a small portion of the genu) and interhemispheric connection by posterior commissure (white arrow) in T1-weighted (A, B), T2-weighted (C) and FLAIR (D) sequences. Putamina, anterior commissure and hemispheric supratentorial white matter are also thin.

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Ethical statement: Full consent was obtained from the patient's family for the case report. This study was approved by our Ethics Institution.

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