Magnetic resonance neuroimaging in phenylketonuria

This patient was diagnosed at neonatal screening with phenylketonuria, an autosomal recessive inborn error of phenylalanine metabolism. Phenylalanine hydroxylase gene analysis showed compound heterozygosity for two previously described sequence variants (p.R408W, p.S349P) known to be pathogenic for phenylketonuria (van Wegberg et al, 2017). From childhood a low phenylalanine diet was prescribed but was not strictly adhered to because of unpalatability.

In the fourth decade, neurological examination showed lower limb hyperreflexia, sustained bilateral ankle clonus but flexor plantar responses. Magnetic resonance brain imaging showed symmetrical areas of signal hyperintensity around the frontal and posterior horns of the lateral ventricles (*Figure 1*), with hazy white matter in the centrum semiovale, and subtle restricted diffusion on diffusion-weighted imaging.

The findings, in keeping with prior reports (Manara et al, 2009), indicate increased myelin turnover which may reverse with resumption of the exclusion diet. Untreated phenylketonuria may cause intellectual impairment, seizures and motor deficits. **BJHM**

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Figure 1. Axial T2-weighted magnetic resonance brain imaging showing confluent white matter change around the horns of the lateral ventricles, particularly evident posteriorly.