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1q44 MICRODELETION SYNDROME – AN ETIOLOGY OF GDD WHICH STILL HASN'T BEEN FULLY EXPLORED

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Abstract

1q44 microdeletion syndrome is a very rare (less than one in a million cases, according to Orphanet) contiguous gene syndrome caused by the loss of a 6 Mb locus on the long arm of chromosome 1 characterized by seizures, global developmental delay and a specific craniofacial dysmorphism. We aim to review the main features of this syndrome and to present the case of a one-year old girl with a normal family history admitted to our hospital for medical recovery who presented global developmental delay and several congenital abnormalities. Genetic testing detected a 967 kb deletion in the 1q44 region and a 530 kb microduplication in the 14q31.1q31.2 region, the latter encompassing no currently known OMIM genes. Our patient's phenotype overlapped 1q44 microdeletion syndrome. After analysing the 1q44 microdeletion syndrome cases reported up to the present time, we discovered that our patient has additional features, which have never been previously described in this syndrome, namely grey matter heterotopy, bifid hallux and prenatal hydronephrosis. Given the renal, cerebral and skeletal impairment in 1q44 microdeletion syndrome, we believe our findings represent additional, previously unreported features of this very rare genetic syndrome.