Juvenile nephronophthisis on MRI—a potential case of Joubert syndrome?

Roslyn J. Simms · John A. Sayer

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Sir,

We read with interest the case presentation of “End-stage juvenile nephronophthisis on MRI” [1]. We are particularly curious about the child’s clinical presentation with an “abnormal gait” [1].

Nephronophthisis accounts for 6.5% of the children requiring renal replacement therapy in the United Kingdom and may be underdiagnosed [2]. As the authors acknowledge, an important feature of nephronophthisis is the association with extra-renal manifestations that can occur in up to 10% [3]. In their discussion, the authors mention Joubert syndrome, which is characterized by a mid-hindbrain malformation, cerebellar vermis hypoplasia and the diagnostic “molar tooth sign” on cerebral MRI [4]. We wonder whether the “abnormal gait” described in this child could be explained by Joubert syndrome and perhaps imaging with a brain MRI would be helpful?

The MRI of the kidneys clearly demonstrates the corticomedullary cysts characteristic of nephronophthisis/medullary cystic kidney disease [1]. Diagnosis should be confirmed by molecular genetic analysis. In isolated nephronophthisis, mutations in NPHP1 account for 20–25% of cases and can be screened for by polymerase

chain reaction (see <http://www.orpha.net> for a list of laboratories) [5]. Additionally, retinal screening for retinal dystrophy is indicated in all cases of suspected nephronophthisis.

Since nephronophthisis is an autosomal-recessive condition, with little genotype-phenotype correlation, the importance of a family history and reviewing other potentially at-risk relatives should be emphasised.

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R. J. Simms (✉) · J. A. Sayer
Institute of Human Genetics—Nephrology,
International Centre for Life, Newcastle University,
Central Parkway,
Newcastle upon Tyne NE1 3BZ, UK
e-mail: rozsimms@doctors.org.uk