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CLINICAL IMAGE

End-stage juvenile nephronophthisis on MRI

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A 10-year-old boy was referred to our institution from his general practitioner with abnormal gait and severe lower limb weakness. Basic admission blood tests revealed severe renal failure and anaemia. Renal US showed bilateral slightly small-for-age kidneys with generalised increase in echogenicity and loss of normal corticomedullary differentiation. A few small cysts were also identified. Non-contrast-enhanced MRI showed additional small cysts mostly at the corticomedullary junction and in the medulla (Figs. 1 and 2).

The history and radiological appearances are typical of end-stage juvenile nephronophthisis (JN), in this case presenting with weakness secondary to renal osteodystrophy.

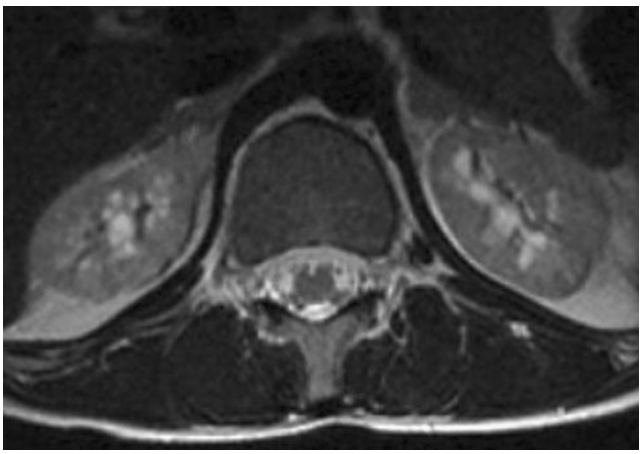

Fig. 1 Axial T2-W MRI of the kidneys

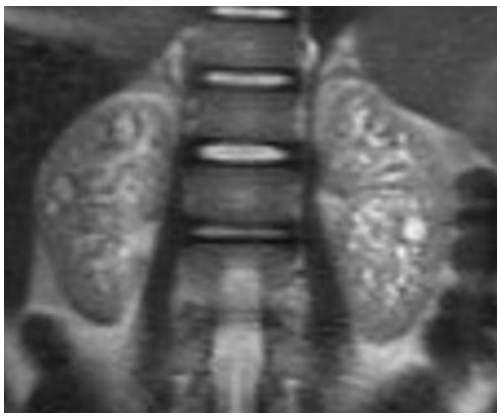

Fig. 2 Coronal T2-W MRI of the kidneys

The above radiological findings have been described as pathognomonic for this condition in a child with severe renal failure. Cysts may not be seen early on in the condition [1]. JN is an autosomal-recessive condition typically presenting in older children as relentless chronic renal failure often undetected until late in the course of the disease. Similar radiological findings are seen in adults suffering from autosomal-dominant medullary cystic disease. The juvenile form has several identified important associations including retinal dystrophy, cerebellar anomalies (Joubert-type anomalies), mental retardation and liver involvement [2].

References

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2. Niaudet P (2004) Nephronophthisis. In: Orphanet encyclopedia. http://www.orpha.net/data/patho/GB/uk-nephro.pdf. Accessed 9th November 2009

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