Correspondence

Nephronophthisis

Sir,

We read with interest the paper by Robins et al. (Archives, 1976, 51, 799) reporting a child with nephronophthisis and liver fibrosis. This observation reinforces the feeling that such association is not fortuitous, as we stated when publishing a similar case (Proesmans et al., 1975) shortly after the original description by Boichis et al. (1973). Our patient also had tapetoretinal degeneration, a condition frequently found in children with nephronophthisis and generally referred to as the Senior syndrome. The ophthalmological findings in our patient have been described in a separate paper (Stanescu et al., 1976), where we stressed the value of the electroretinogram (ERG) in diagnosing retinal diseases. Robins et al. state, 'There was no evidence of retinal changes in the present case'. We wonder if an ERG was performed. The authors did find the skeletal anomalies reported by Mainzer et al. (1970) in 2 sibs with nephronophthisis. Our patient did not have these specific bone lesions.

Several questions arise. Should we go on creating eponyms now that Senior syndrome is a widely used term, and a series of more or less specific anomalies in other organs are known to occur in association with chronic tubulointerstitial nephritis? Is it reasonable to introduce the following ones: Mainzer syndrome (nephronophthisis associated with conical epiphyses), Boichis syndrome (nephronophthisis with liver fibrosis), Senior-Boichis syndrome in the case we described, and Mainzer-Boichis syndrome in the case of Robins et al.? We think the answer is no. Besides it is highly probable that in the past many cases of nephronophthisis were incompletely examined or reported. We are aware of two examples. In the original paper by Fanconi et al. (1951) it is mentioned very briefly that in at least one of the two families reported mental retardation was present in the affected sibs. This point has never been stressed. In the case we reported there was mild ataxia, which was not further elaborated on and therefore not mentioned in the paper. Finally, what is nephronophthisis? Is it a single entity or just one of many forms of chronic tubulointerstitial nephropathy?

In order to answer these questions, all patients with chronic interstitial renal disease should be studied as completely as possible, examining the eyes (including ERG), the central nervous system (including intellectual performance), the skeleton (with special attention to phalanges and femora), and the liver (for fibrosis).

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References


Dr. Robins comments:

We agree with the points made by Drs. Proesmans and Van Damme. Juvenile nephronophthisis is possibly not a disease entity and we were therefore careful to state that the features in our patient were 'consistent' with this condition. Adopting eponymous titles is unlikely to serve any useful purpose but it is hoped our knowledge will increase if more patients with the disease are investigated along the lines suggested (though never at the expense of a particular child's welfare). An electroretinogram was not performed on our patient and our statement that his retinae were normal was based on clinical examination.

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Transverse myelitis—association with Coxsackie B3 infection?

Sir,

We report the possible causal association of transverse myelitis and Coxsackie B3 virus infection in a 7-year-old girl. Her illness started with frontal headache, vomiting, and pyrexia, and as these symptoms were persisting she was admitted to hospital on day 9 of the illness, though clinical examination was essentially negative. On day 13 she was first found to have increased tone in both legs, with exaggerated tendon reflexes and bilateral extensor plantar responses. There was then a rapid progression of symptoms, with greatly diminishing power in the legs,