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 detail 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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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 Cocksackie B₃ infection?

Sir,

We report the possible causal association of transverse myelitis and Cocksackie 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,

retention of urine with overflow, faecal incontinence, drowsiness, and neck stiffness. 17 days from the onset loss of vibration sense, diminution in joint position sensation, and a qualitative loss of sensation to pinprick had developed, involving both legs and extending to the area supplied by the 8th cervical spinal segment. Some return of power in the legs was noted on day 19 of illness, and over the next week there was a steady improvement. 6 weeks later she had only slight weakness of the legs, but after 6 months there were no abnormal signs, and her school reported her academic ability apparently unimpaired.

Lumbar puncture after admission produced clear cerebrospinal fluid under normal pressure containing $15/\text{mm}^3$ white cells, mainly mononuclear, protein 0.36 g/l , and glucose 2.5 mmol/l ($45 \text{ mg}/100 \text{ ml}$), which was sterile on culture. A further tap a few days later showed a slightly higher white cell count ($46/\text{mm}^3$, 90% lymphocytes) but a lower protein. A marked neutrophil leucocytosis (total white cell count $41.0 \times 10^9/\text{l}$ ($41000/\text{mm}^3$) with 87% polymorphs) was present on admission and persisted for 4 weeks. A thorough and repeated search for bacterial infection including a bone scan was negative. Acute and convalescent antibody titres done in parallel did not suggest infection with *Brucella*, Epstein-Barr virus, *Rickettsia*, adenovirus, mumps, cytomegalovirus, measles, herpesvirus, *Toxoplasma*, *Leptospira*, *Staphylococcus*, or *Salmonella typhi* and *paratyphi*. Coxsackie antibody titres also were normal except for B_2 which showed a raised titre on admission (1:1024), falling to 1:256 by day 19 of illness. Viral cultures were negative.

We feel this very high Coxsackie B_2 titre suggests that the illness was caused by this organism, and we cannot find any published reports of this combination, though transverse myelitis has been described in association with B_4 virus (Dery *et al.*, 1974). The persistent neutrophil leucocytosis, which was a worrying feature of the illness, does not seem to have been a feature of the rare cases of transverse myelitis with other viral aetiologies.

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Reference

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Maternal diabetes and congenital malformation

Sir,

May I add to the experience of Drs. Day and Insley detailed in their paper on this subject (*Archives*, 1976, **51**, 935), as it is very similar to my own (Komrower, 1961). I followed 213 consecutive pregnancies where all the mothers had received insulin therapy. There were 20

deaths before the 28th week (14 less than 20 weeks), 16 stillbirths and 20 neonatal deaths. I was able to review 182 children on more than one occasion, some for more than 10 years, and by good chance I adopted similar definitions to Day and Insley for major and minor malformations.

There were 19 significant congenital abnormalities: 5 in stillbirths, 3 in neonatal deaths, and 11 in living children. The system involvement was similar to that described by the Birmingham authors—cardiovascular 5, skeletal 5, urogenital 4, oesophageal atresia 1, central nervous system 2 (one child had both anencephaly and multiple skeletal deformities), and 3 had large haemangiomas. The incidence was 9.75% compared with a hospital figure of 3.2% for a 2-year period during the study (the latter results were obtained from our monthly neonatal report). Unfortunately, I do not have the ages of the mothers concerned but the duration of their diabetes varied between 6 months and 10 years. This study took place before our present experience in the treatment of hyaline membrane disease but even so one was able to show that a diabetic woman reaching the 28th week of her pregnancy had a 75% chance of producing a healthy baby without a congenital abnormality.

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Reference

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Fibrosing alveolitis

Sir,

Dr. Hewitt and his colleagues, in their paper on fibrosing alveolitis in infancy and childhood (*Archives*, 1977, **52**, 22), state that all but 2 of their 10 children improved when given corticosteroids, and in their summary they further state that in children fibrosing alveolitis is almost always a corticosteroid-responsive disease. This being the case, I should be interested to learn why 5 of their patients died of the disease, including 4 shown in Table 8 to be steroid-responsive.

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Prof. D. Hull and Dr. C. J. Hewitt comment:

We anticipated that some readers of the article might raise questions that could be more appropriately answered by full detailed case histories. We would be prepared to send these direct to any interested clinician.