

CASE 1994 - 9

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Clinical History: This infant was the product of a normal, 36 week pregnancy in 38 year old gravida ii woman. Amniocentesis and fetal karyotyping for advanced maternal age showed a normal fetal karyotype. Apgars 6, 8 and 9. Birth weight 2270 gms. Length 70 cm (90%ile) and head circumference 25 cm (1½ cm below 10%ile). Referred at 16 days of age for dysmorphic features and congenital nephrotic syndrome. Dysmorphic features were ? posteriorly rotated ears, small palpebral fissures with microphthalmia, high-arched palate, micrognathia, wide-spaced nipples, flexion deformities of MPJ of thumbs, (also described as arachnoidactyly). The renal problems were edema, including orbital puffiness, proteinuria, and impaired renal function. The kidneys were palpable and ultrasound showed enlarged kidneys. Head ultrasound showed that the contour of both cerebral hemispheres appeared smooth and simple with lack of sulcal formation, and the Sylvian fissure also appeared simple. Over the last 15 days of life hemoglobin fell from 158-172 to 91 g/L, serum sodium potassium and calcium tended to be slightly under the lower limit of normal, urea varied from 14.6 - 15.0 mmol/L, creatinine rose from 169 to 269 umol/L. Total protein was 32 g/L; serum albumin ranged from 19-25 g/L and beta and gamma globulins were also low. Urine protein was more than 5.00 g/L. Alkaline phosphatase ranged from 93-55 U/L. Investigations for various viral and bacteriological agents were negative. The EEG was severely abnormal.

The infant was treated with many infusions of albumin. A feeding jejunostomy was performed at 23 days of age because of hiatus hernia, gastric reflux and esophageal motility problems in the hopes that this would allow the baby to be sent home to die, the diagnosis having been made clinically. After the operation he required intermittent positive pressure ventilation. Seizure activity was seen. He became oliguric, and his condition deteriorated, with gasping respiration. He was extubated and died at 31 days.

Necropsy findings: Failure to thrive (weight 2890, normal 3634 g) Brain weight 172.3 g (normal 431 +/- 117.7). Pachygyria, kidneys showed glomerulosclerosis, tubular ectasia, renal hyperlobation. Other findings: hiatus hernia, thyroid dysplasia, adrenal hypoplasia, facial dysmorphism (micrognathia, posteriorly rotated ears, high arched palate), flexion deformity of thumbs, wide-spaced nipples, generalized edema with pleural effusions and ascites, acute pneumonia, hepatic cholestasis with ductular proliferation.

Material submitted: 1 Kodachrome of brain.
1 H & E section of frontal lobe.

Points for discussion: 1. Diagnosis
2. Pathogenesis

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