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CLINICAL QUIZ

Congenital nephrotic syndrome with dysmorphic features and death in early infancy: Questions

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Case summary

The baby boy was the second child of Austrian parents who reported no consanguinity. His twin sibling had died at 10 weeks of gestation. Birth occurred after 36 weeks; weight, length and head circumference at birth were 2,090 g [<3rd percentile (2.5 kg)], 43 cm [<3rd percentile (46.3 cm)] and 31 cm [<3rd percentile (32.1 cm)], respectively. The Apgar score was 6/8/9. Due to respiratory distress, the newborn was transferred to the neonatal intensive care unit. He showed multiple petechiae, inverted nipples and scrotal and penile oedema. Facial dysmorphisms, such as a broad nasal bridge and distinctly low-set ears, as well as small eyes with pronounced hypertelorism were present. The right foot presented as pes supinatus with digitus quintus subductus.

The patient suffered from congenital nephrotic syndrome (CNS) with generalised oedema, severe proteinuria (645 mg/dl), hypoalbuminemia (20.6 g/l) and low serum immunoglobulin G [IgG; on day 27: <40 mg/dl (normal range 660–1,750 mg/dl)]. He was treated with daily protein substitutions (6 ml/kg human albumin 20 %) and IgG substitutions. High blood pressure developed. Attempts to treat the CNS with corticoids were unsuccessful.

Levels of thyroid hormones were normal at 14 days of age, but subsequently decreased: TSH 0.44 (reference 0.72–11.00) μU/ml, free T4 0.52 (reference 0.89–2.20) ng/dl and free T3 1.05 (reference 1.95–6.04) pg/ml. Thrombocytes were low (70,000/μl).

Seizures started at 52 days of age, and electroencephalography revealed a burst suppression pattern in the fronto-central region of the right hemisphere. Due to muscular hypotonia a biopsy of the quadriceps femoris was performed, showing a predominance of type 2 muscle fibres (approx. 75 % of all fibres) and relatively small type 1 muscle fibres (about 30 % smaller than type 2 fibres).

At 6 weeks of age, the patient developed pneumonia and sepsis. Anuria occurred, and the boy died from kidney and respiratory failure 1 week later.

Questions

1. What is the differential diagnosis for the underlying disorder?
2. What test result should be obtained to establish the diagnosis?

The answers to these questions can be found at http://dx.doi.org/10.1007/s00467-015-3070-1.

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