Eur J Pediatr (2003) 162: 559–561
DOI 10.1007/s00431-003-1239-2

YOUR DIAGNOSIS

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An 11-month-old boy with psychomotor regression and auto-aggressive behaviour

Received: 6 March 2003 / Accepted: 2 April 2003 / Published online: 16 May 2003

Clinical information

An 11-month-old Swiss boy was brought to his paediatrician. He had been in good health and had developed normally until then, but his parents mentioned that over the previous 2 weeks, the child no longer laughed or played, was becoming more and more restless, and slept only 1 to 2 h a night. He was no longer able to crawl and to stand up, and he had lost weight. Clinical examination revealed swollen hands and feet with skin desquamation, axial hypotonia and brisk reflexes. The child sweated profusely, refused to crawl or stand, showed stereotypic movements of the hands (kneading) and repeatedly bit objects or his own hands.

Upon admission to the regional hospital, blood cell count, electrolytes, blood gas analysis, thyroid hormones and cerebrospinal fluid were normal. Liver and kidney parameters were normal, but plasma LDH and CK were increased to 724 U/l (normal < 296 U/l) and to 270 U/l (normal < 234 U/l), respectively. EEG, chest X-ray film, abdominal sonography, cranial CT scan and a muscle biopsy were all normal. Differential diagnosis at that point included a viral infection, neuroborreliosis, and coeliac disease, but laboratory investigations remained inconclusive.

Two further weeks later, the symptoms persisted and the child was referred to a secondary centre. Clinical findings were unchanged; routine laboratory studies were again normal. EEG, brain MRI, and investigations for a metabolic disorder (ammonia, uric acid, amino acids, organic acids, acylcarnitines and mucopolysaccharides) were normal. The diagnosis remained obscure. The child was referred for further evaluation of severe psychomotor regression with autistic features of unknown aetiology.

The child was admitted to our hospital at age 14 months. Length and weight, that had been at P50 at age 11 months, had declined to P3–10. The child appeared exhausted and irritable and sweated profusely. The hands and feet were swollen, erythematous and tender (Fig. 1). A fine maculo-papular exanthema was present on the trunk. There was muscular hypotrophy with peripheral oedema and axial hypotonia. Physical findings included tachycardia (160–180/min) and arterial hypertension (140/80 mmHg). The boy refused to stand, but would sit in a crouched position, had a sad and apathetic look, took little interest in his surroundings and would occasionally bite his hands and feet. A review of a videotape obtained during the first hospitalisation several weeks earlier showed that all features had been even more marked at that time.

Abdominal sonography showed no signs of a tumour or renal disease; chest radiography and MIBG-scintigraphy showed no signs of a tumour. Echocardiography showed no signs of cardiomyopathy. An ophthalmological examination did not reveal corneal or lenticular abnormalities. An afebrile seizure occurred during hospitalisation: EEG and cerebral spinal fluid were again normal. Arterial hypertension and tachycardia had a good response to beta-blockers. Thyroid hormones were normal, but urinary excretion of epinephrine (epinephrine/creatinine: 86 nmol/mmol; normal < 25 nmol/mmol), norepinephrine (norepinephrine/ creatinine: 263 nmol/mmol; normal < 55 nmol/mmol), dopamine (dopamine/creatinine 1192 nmol/mmol; normal < 590 nmol/mmol) and HMA and VMA (HMA/creatinine 6.9 μmol/mmol; normal < 4.7 μmol/mmol) were elevated.

Because of developmental regression, muscular hypotonia, anorexia, and auto-aggressive behaviour with biting of feet and fingers, Lesh-Nyhan syndrome was considered but excluded by normal levels of uric acid and purine pattern in plasma and urine. Fabry disease was considered because of hypertension and pain in hands and feet but ruled out by normal α-galactosidase activity in leukocytes. Finally, a specific question to the child's parents allowed a presumptive diagnosis.

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