

Autopsy brain

The patient was a 23 month old male child

was the product of a normal, full term pregnancy, and uncomplicated delivery. At the age of six weeks, he began to suffer repeated respiratory tract infections and was reported to have episodes of fever, rectal bleeding, thrombocytopenia, and bone marrow hyperplasia at about the same time. Exploratory laparotomy was negative. At the age of seven months, he weighed 18 pounds but further weight gain did not occur in spite of all therapeutic efforts. Development was most advanced at eight months when he was able to sit up and hold his bottle; however, at twelve months he was unable to hold up his head with persistence and at twenty-one months he was unable to sit or roll over. Extensive investigations were repeatedly carried out all of which were normal except for a transient low hemoglobin, a moderate acidosis, and a mild hyperchloremia. During the last year of life, the outstanding clinical manifestations had been generalized, moderate, muscular wasting and progressive weakness. The neurologic findings were considered negative except for moderately hypoactive reflexes, "strange movements of the eyes", generalized hypotonia, and retarded development. The nutritional state was considered to have been maintained "amazingly well despite the progressive muscular wasting". Spinal fluid examination performed at age 12 months and 17 months was normal except for a protein of 80 mg. % and 65 mg. % respectively. Skull and chest X-rays were normal and subdural taps produced no fluid. Electrocardiogram and electromyogram showed no significant abnormalities. No positive diagnosis was ever arrived at though the clinical diagnostic impression was "amyotonia congenita." The patient's nutritional state was maintained by tube feedings and he was also given a trial of cortisone without apparent benefit.