Ultrastructural Studies of Type II Fucosidosis

B. Porfiri¹, R. Ricci², D. Seminara³, and G. Segni³

¹ Istituto di Istologia ed Embriologia Generale, Università Cattolica del S. Cuore, Facoltà di Medicina e Chirurgia, Via Pinetta Sachetti 644, I-00168 Roma, Italy
² Istituto di Biologia e Zoologia Generale, Università Cattolica del S. Cuore, Facoltà di Medicina e Chirurgia, Roma, ed Istituto di Istologia ed Embriologia, Facoltà di Scienze di Camerino, Italy
³ Istituto di Clinica Pediatrica, Università Cattolica del S. Cuore, Facoltà di Medicina e Chirurgia, I-00168 Roma, Italy

Summary. Type II fucosidosis in an autosomal recessive disease. The paper presents a case of a patient with α-L-fucosidase of whom a skin specimen was examined under the electron microscope. Storage material was observed mainly in endothelial cells of blood capillaries and Schwann cells surrounding small peripheral nerves of papillary dermis. Within both cells two different kinds of inclusions were revealed: (1) clear vacuoles and (2) dense bodies with an internal structure prevalently lamellar.

All these ultrastructural alterations were observed long before the appearance of clinically defined angiokeratoma at cutaneous level. Hence, they present the same alteration found in the absence of angiokeratoma in type I fucosidosis.

Key words: Microscopy, electron — Skin — Type II fucosidosis


Schlüsselwörter: Mikroskopie, Elektronen- — Haut — Fucosidosis Typ II

Offprint requests to. B. Porfiri, MD (address see above)
Fucosidosis, an autosomal recessive inborn error of metabolism, is determined by the deficiency of the activity of lysosomal hydrolase α-L-fucosidase [1—3].

Two types of fucosidosis were identified with different clinical findings. Type I fucosidosis [4] is characterized by a severe psychomotor involvement starting in the first year of age, severe progressive cerebral degeneration, gradual loss of muscle strength, spasticity, and finally decerebrate rigidity. Death occurs in the first 5 years. Type II fucosidosis [5, 6] is characterized by a late onset of psychomotor regression at the age of about 15—17 months, angiokeratoma diffusum, and a later age of death. Patients with type II fucosidosis may survive into early childhood.

Skin lesions should hence be characteristic clinical signs that distinguish type II from type I fucosidosis. However, little electron-microscopic research is available as yet [7—9].

We present an ultrastructural study carried out on a skin biopsy from a patient with type II fucosidosis.

Case Report

A 4.5-year-old girl was admitted from the Neurological Clinic to the Pediatric Clinic of the Università Cattolica of Roma on September 14, 1977, for psychomotor regression. She was the second child of consanguineous (first cousins) Italian healthy parents whose first child was a normal girl. She presented a normal neonatal growth and development until the age of 10—12 months. Afterwards psychomotor regression with mild deafness appeared.

Facial feature does not show the typical gargoyl aspect, except for thick tongue and widely spaced teeth. Obliged posture of the head toward upright with marked cervical lordosis is evident (Fig. 1).

Thorax and cardiac examination was normal. Abdomen was protruding with mild heptosplenomegaly.

Mental development was considerably delayed and, at the age of 4.5 years, corresponded to that of a 10—12-month-old infant. Neurologic examination revealed a mild ocular dispareisis and left hemiparesis. Electron-encephalic examination revealed modifications of irritable origin, firstly in the forehead region. The walking was oriented to the right. Sensibility was normal and sphincteric function controlled. She presented a severe deafness and her language was compromised since she was unable to articulate phonemes.

Ocular examination showed a superficial corneal dystrophy more evident on the left. A defective implantation of teeth with high abrasion of masticatory surfaces was observed.

Dermatologic examination did not reveal any skin alteration (16/9/77).

Routine laboratory studies were normal. Urinary excretion of uronic acid was normal. Cetyl peritidium precipitation (CPC) of urine, followed by chromatographic analysis [10], revealed an increased amount of keratan-sulfate, in the fraction eluted with 2 and 3 M NaCl, as compared to normal controls.

Blood group is A Rh+, as in the other members of the family.

The girl presented an increased salinity of sweat (75 mEq/l).

X-ray findings showed a mild delayed skeletal maturation at the level of metacarpals; vertebral bodies resulted somewhat morphologically altered; the femoral neck appeared longer and thinner than normal.

After 20 days of hospitalization the patient was discharged. At present, she is 6.5 years old and presents steady general conditions although neurological symptoms are moderately worsened.

A dermatologic examination performed 2 months ago (September 1979) revealed a beginning angiokeratoma characterized by clusters of dark red, punctate lesions around the genitalia and on palms and sole.

Enzyme Studies

Assay of enzymatic activity of α-L-fucosidase was performed according to Zielke’s method using p-nitrophenyl-fucoside as substrate (9 mM) [11]. Lymphocytes from peripheral blood were purified by