Late-Onset Type Cystinosis: Report of a Case

EN LIN, KUO-HSING CHEN, CHING-SHIANG CHI, SHI-HUANG LEE*,
WILLIAN L. HO **, and KWANG-JEN HSIAO***

A 10-year-old girl was diagnosed as having cystinosis at the age of 7. She was found to have growth retardation and bow-legs. X-ray of the long bones revealed rickets. Prominent glucosuria and aminoaciduria with a borderline glomerular filtration rate were noted. A slit-lamp examination of the eyes showed refractile tinsel-like crystals deposited in the cornea and conjunctiva but fundoscopic examination gave no abnormal retinal findings. Cystine crystals were demonstrated from conjunctival biopsy tissue by both light and electron microscopy. Skin fibroblast cystine-uptake assay proved to be a case of homozygous cystinosis. we considered this case to be a late-onset form of cystinosis with clinical symptoms presenting at the age of 16 months. Clinical manifestations included renal tubular dysfunction, slowly progressive renal impairment, and characteristic corneal and conjunctival cystine deposits without retinopathy.

Key words: *cystinosis, late onset form cystinosis, refractile tinsel-like crystals*

(*J Formosan Med Assoc* 1987; 86:1307-11)

Cystinosis is an autosomal recessive disorder in which the disulfide amino acid cystine accumulates intracellularly within lysosomes in many organ tissues such as the cornea, conjunctiva, bone marrow, lymph nodes, and leukocytes [1,2]. The etiology of this biochemical defect is obscure. In this report, we describe a Taiwanese girl with cystinosis, who started chinical sympt oms of rickets and Fanconi's syndrome in early childhood. We will also briefly review the literature.

CASE REPORT

A 10-year-old girl was diagnosed to have cystinosis at the age of 6 years and 9 months. She was brought to our clinic 3 years ago for growth retardation and bow-leg. Since 16 months of age, she had failed to thrive, and had glucosuria and mild proteinuria. There was neither hereditary disease nor consanguineous mating in her family. Slit-lamp examination of her family members, including her sisters and brothers, did not demonstrate tinsel-like cystine crystals deposited in the cornea or conjunctiva. Photophobia or skin hypopigmentation was not noticed.

Fig. 1. *Slit-lamp photograph of cornea showing numerous refractile cystine crystals.*

Departments of Pediatrics, Ophthalmology,* and Pathology.** Veterans General Hospital-Taichung, Clinical Biochemistry Research Laboratory, Department of Medical Research, Veterans General Hospital-Taipei***, Taiwan, R.O.C.
Received: May 19, 1987. Revised: August 10, 1987. Accepted: September 8, 1987.
Address reprint requests to: Dr.En Lin, 160, Section III, Chung-Kang Rd, Taichung, Taiwan