Fingerprint Profiles in Lymphocytic Vacuoles of Mucopolysaccharidoses I-H, II, III-A, and III-B

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Summary. Fingerprint (FP) profiles in vacuolated lymphocytes of mucopolysaccharidoses I-H, II, III-A, and III-B are a numerically rare, but possibly consistent finding as they have not been seen in vacuolated lymphocytes of other non-neuronal lipofuscinosis (NCL) lysosomal diseases. Their nosologic significance is not clear, but they may be as non-specific as tubular inclusions in lymphocytes and they are identical to those FP profiles seen in juvenile NCL.

Key words: Fingerprint profiles — Vacuolated lymphocytes — MPS I-H, II, and III — Juvenile NCL

Introduction

Vacuolization of lymphocytes appears in a number of lysosomal diseases, such as mucopolysaccharidoses (MPS), mucolipidoses, GM1-gangliosidoses and juvenile neuronal ceroid-lipofuscinosis (NCL). In NCL, membrane-bound lysosomal compartments of lymphocytes are also filled with granular material, curvilinear profiles, or fingerprint profiles (Schuurmans, Stekhoven et al. 1976; Ikeda and Goebel 1979). Fingerprint (FP) profiles are particularly conspicuous in lymphocytes of juvenile NCL (Schwendemann 1976; Baumann and Markesbery 1978), where they are often embedded in a clear vacuolar matrix. To recognize such FP profiles, or fingerprint profiles (Schuurmans Stekhoven et al. 1976) in other non-NCL lysosomal disorders marked by lymphocytic vacuoles.

It is the purpose of this communication to present non-NCL intravacuolar FP profiles in lymphocytes of MPS I-H, II, III-A, and III-B.

Results

Among a large number of lymphocyte preparations, screened for the following lysosomal disorders diagnosed by clinical, morphological and biochemical criteria, MPS I-H, II, III-A, III-B, IV-A, VI-A, and VI-B; GM1-gangliosidoses I and II; NCL, infantile, late infantile, and juvenile types; Krabbe’s and metachromatic leukodystrophies; Gaucher’s disease; and type II glycogenosis, intravacuolar FP profiles in lymphocytes (Fig. 1) were only detected in one male patient afflicted with MPS I-H, two male patients afflicted with MPS II, one male patient afflicted with MPS III-A, one female patient afflicted with MPS III-B, and those afflicted with juvenile NCL. Whereas empty vacuoles were rather numerous in lymphocytes of these MPS patients, vacuoles that contained the FP profiles appeared scanty, usually there were not more than one or two in the entire lymphocyte preparation. Not infrequently, such FP profiles were attached to lipid droplets inside the MPS lymphocytic vacuoles (Fig. 1). The thick leaflets measured between 5 and 5.7 nm, the light leaflets between 2.3 and 3.4 nm, and there were no appreciable differences in measurements between FP profiles in MPS lymphocytic vacuoles and FP profiles in NCL lymphocytic vacuoles.

Comment

Although FP profiles are regarded as an ultrastructural hallmark of NCL, especially of the juvenile type (Zeman et al. 1970; Goebel et al. 1979), they rarely may occur in other disorders (Goebel and Schulz 1979) and apparently also in MPS lymphocytes. Such intravacuolar FP profiles have previously been reported in lymphocytes of MPS II and III; they had been rather scant in number (Markesbery et al. 1980). We are able to confirm these findings and add that MPS I-H as well as both types of MPS III-A and III-B, may show FP profiles in lymphocytic vacuoles.

The pathogenetic significance of these FP profiles in MPS I-H, II, III-A, and III-B vacuolated lymphocytes is unclear. They might represent a non-specific finding as the tubular inclusions seen in many lymphocytes of lysosomal and non-lysosomal disorders.
Lymphocytes in juvenile NCL are marked by aggregates of membrane-bound vacuoles which contain FP profiles. Such FP profiles vary in number and amount within these vacuoles, but they are sometimes so scant that empty vacuoles predominate numerically in such lymphocytes of juvenile NCL. Then, it is almost impossible to distinguish diagnostically between vacuolated lymphocytes of juvenile NCL and vacuolated lymphocytes of non-NCL lysosomal disorders. It is also unknown whether lymphocytic vacuoles in juvenile NCL contain a similar chemical compound or compounds as the lymphocytic vacuoles in other non-NCL lysosomal disorders, especially the MPS I-H, II, III-A, and III-B.

References