Brief Communication

J Mucopolysacch Rare Dis 2016;2(1):31
http://dx.doi.org/10.19125/jmrd.2016.2.1.31
pISSN 2465-8936 · eISSN 2465-9452

JMRD
Journal of Mucopolysaccharidosis and Rare Diseases

Mucolipidosis Type II in Vietnam

Vu Chi Dung

Department of Medical Genetics, Metabolism and Endocrinology, Vietnam National Children's Hospital, Hanoi, Vietnam

Purpose: To describle clinical features and enzyme activity of Vietnamese patients with Mucolipidosis type II.
Methods: Clinical features, laboratory and plasma lysosom enzyme activity by 4 MU-Fluorometric assay was studied from 2014–2015 at the Northern referral center of Pediatrics – National Children's Hospital.
Results: 16 cases (7 girls and 9 boys) were diagnosed with I-cell bases on clinical symptoms and enzyme activities studies. Diagnosis age was 5.93±4.28 years, onset age was recognised from birth to 4 years (median 1.25) with the feature of joint stiffness and bone deformation. All cases presented with the feature of joint stiffness, chest deformation and kyphoscoliosis; Fifteen cases (93.7%) had coarse facial features. No patients had hepatosplenomegaly on abdominal ultrasound, 5/15 patients had heart valves disease. Enzyme assay showed α-Hexosaminidase of 1,885.9±338.7 (nmol/mg plasma/17 hrs), α-Iduronate sulfatase of 4,534.8±1,062.9 nmol/mg plasma/4 hrs).
Conclusion: Mucolipidosis II seriously affected the life of the patients with skeletal deformities, contractures develop in all joints and cardiac involvement.

Keywords: Mucolipidosis type II, I-cell disease

I-cell disease (Mucolipidosis II) is a rare lysosomal storage disorder caused by the deficiency of N-acetylglucosamine-l-phosphotransferase, an enzyme that transfers phosphate groups into oligosaccharide units of lysosomal enzyme precursors. Due to the absence of transferase activity, the common phosphomannosyl recognition marker of acid hydrolases is not generated, and the enzymes are not targeted to the lysosomes I. As a consequence the enzymes are secreted into the extracellular space, and high activities can be found in the serum, cerebrospinal fluid and urine of the patients, whereas inside the cells (fibroblasts) the enzyme levels are considerably reduced. Mucolipidosis II is also known as I-cell disease because of the coarse granular cytoplasmic inclusions seen in cultured skin fibroblasts which are large lysosomes containing heterogeneous material.

Received April 12, 2016; **Revised** April 22, 2016; **Accepted** May 10, 2016
Correspondence to: Vu Chi Dung
Department of Medical Genetics, Metabolism and Endocrinology, Vietnam National Children's Hospital, No. 18/879 La Thanh Road, Dong Da District, Hanoi, Vietnam
Tel: +84-4-37534548, Fax: +84-4-62738573, E-mail: dungvu@nhp.org.vn