



POSTER PRESENTATION

Open Access

Mutations of *ABCD1* gene and phenotype of Vietnamese patients with X-linked adrenoleukodystrophy (X-ALD)

Vu Chi Dung^{1*}, Nobuyuki Shimozawa², Nguyen Ngoc Khanh¹, Bui Phuong Thao¹, Can Thi Bich Ngoc¹, Nguyen Phu Dat¹, Nguyen Thi Hoan¹

From 7th APPEs Biennial Scientific Meeting
Nusa Dua, Bali. 14-17 November 2012

X-linked adrenoleukodystrophy (X-ALD) is caused by a defect in the gene *ABCD1*, which maps to Xq28 and codes for a peroxisomal membrane protein that is a member of the ATP-binding cassette transporter superfamily. X-ALD is panethnic and affects approximately 1:20,000 males. This disease characterized by progressive neurologic dysfunction, occasionally associated with adrenal insufficiency.

We aim to describe the clinical, laboratory and cerebral MRI characteristics of Vietnamese patients with X-ALD and to identify mutations of *ABCD1* in these cases. Clinical features, biochemical finding and cerebral MRI lesions of 9 cases from 7 unrelated families were studied. Genomic DNA from these patients was extracted using standard procedures from the peripheral blood leukocytes. Mutation analysis of *ABCD1* was performed using Polymerase chain reaction (PCR) and DNA direct sequencing.

Among these patients, two families had two children with X-ALD, the others were unrelated but one case had family history of X-ALD (his maternal grand mother has a sister whose two sons having paralysis for more than 1 year, hyper-pigmentation and died at the age of 7 and 12 yrs). Endocrinology symptoms of adrenal insufficiency were observed in 8/9 cases; 7/9 cases showed neurological symptoms of cerebral ALD or adrenomyeloneuropathy; 2/9 cases had only symptoms of chronic adrenal insufficiency and no neurological symptoms until 12 and 5 years of age, respectively. 8/9 cases had serum cortisole and ACTH measured confirmed adrenal insufficiency. 8/8 cases showed increased plasma VLCFA. Neuroimaging studies (cerebral MRI) showed classical posterior pattern in 7 cases who had neurological symptoms

and normal pattern in 2 cases without neurological manifestations. We identified 7 different mutations of *ABCD1* in 9 patients. Of which, four novel mutations [c.1202G>T (p.Arg401Trp); c.1208T>A (p.Met403Lys); IVS8+28-551bp del; and the extent of deletion included between IVS1+505 and IVS2+1501, containing whole the exon 2 (4243bp), plus insertion of 79bp from BAP31 and 8bp from unknown origin in this deleted region] were identified in four unrelated patients with neurological symptoms. The reported mutation c.1628C>T (p.Pro543Leu) was identified in two cases (sibling: elder had no neurological symptoms and younger had progressive neurological disability). The reported mutation c.1553G>A (p.Arg518Gln) was found in a boy without neurological symptoms at 5 years of age. The reported mutation c.1552 C>T (p.Arg518Trp) was identified in two cases (sibling: both have adrenal insufficiency and neurological symptoms).

For the first time, mutations in *ABCD1* are identified in X-ALD Vietnamese patients. Despite many mutations having been identified in patients with these clinical phenotypes, the genotype-phenotype correlations have not been clarified.

Authors' details

¹Vietnam National Hospital of Pediatrics, Hanoi, Vietnam. ²Division of Genomics Research, Life Science Research Center, Gifu University, Department of Pediatrics, Gifu University School of Medicine, Gifu, Japan.

Published: 3 October 2013

doi:10.1186/1687-9856-2013-S1-P127

Cite this article as: Dung et al.: Mutations of *ABCD1* gene and phenotype of Vietnamese patients with X-linked adrenoleukodystrophy (X-ALD). *International Journal of Pediatric Endocrinology* 2013 **2013**(Suppl 1):P127.