Newborn Screening, Inborn Errors of Metabolism

AB067. X-linked adrenoleukodystrophy: Phenotype and genotype in Vietnamese patients

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Background: 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. This disease characterized by progressive neurologic dysfunction, occasionally associated with adrenal insufficiency. Objective is to identify phenotype and genotype in Vietnamese patients with X-ALD.

Methods: Genomic DNA from 20 Vietnamese patients from 18 unrelated families 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.

Results: We identified 17 different mutations of *ABCD1* in 20 patients including missense mutations (2/17), deletion (4/17), frameshift mutation (1/17) and splice site mutation (1/17). Of which, six novel mutations including c.1202G>T (p.Arg401Trp); c.1208T>A (p.Met403Lys); IVS8+28-551bp del; c.1668G>C (p.Q556H); c.292_296delTCGGC (p.S98RfsX95); 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 six unrelated patients. Eleven reported mutations including c.796G>A (p.Gly266Arg); c.1628C>T (p.Pro543Leu); c.1553G>A (p.Arg518Gln); c.1552 C>T (p.Arg518Trp); c.854G>C (p.R285P); c.1825G>A (p.E609K); c.1415_1416delAG (p.Q472RfsX83) and c.46-53del insG, c.1553G>A (p.Arg518Gln), c.1946-1947insA (p.Asp649fsX733), c.1978C>T (p.Arg660Trp) were identified in 14 patients from 12 families. Most of patients (17/20) presented cerebral ALD type with/without adrenal insufficiency and only 3 patients presented Addison type.

Conclusions: Mutation analysis of *ABCD1* gene helped confirmation of diagnosis of X-ALD, genetic counselling and prenatal diagnosis but could not be used to predict the specific phenotype of X-ALD.

Keywords: X-linked adrenoleukodystrophy (X-ALD); *ABCD1* mutations

doi: 10.21037/atm.2017.s067

Cite this abstract as: Nguyen KN, Nguyen HT, Can NT, Bui TP, Nobuyuki S, Vu HA, Do MT, Vu DC. X-linked adrenoleukodystrophy: Phenotype and genotype in Vietnamese patients. Ann Transl Med 2017;5(Suppl 2):AB067. doi: 10.21037/atm.2017.s067

