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Novel exon nucleotide deletion causes adrenoleukodystrophy in a Brazilian family

E.R. Valadares[2,4]*, A.L.C. Trindade[1]*, L.R. Oliveira[3,4], R.R. Arantes[4], M.V. Daker[4], B.M. Viana[4], V.G. Haase[5], L.B. Jardim[6], G.C. Lopes[1] and A.L.B. Godard[1]

[1]Laboratório de Genética Animal e Humana,
Instituto de Ciências Biológicas, Departamento de Biologia Geral,
Universidade Federal de Minas Gerais, Belo Horizonte, MG, Brasil
[2]Departamento de Propedêutica Complementar,
Faculdade de Medicina, Universidade Federal de Minas Gerais,
Belo Horizonte, MG, Brasil
[3]Departamento de Pediatria, Faculdade de Medicina,
Universidade Federal de Minas Gerais, Belo Horizonte, MG, Brasil
[4]Hospital das Clínicas, Faculdade de Medicina,
Universidade Federal de Minas Gerais, Belo Horizonte, MG, Brasil
[5]Departamento de Psicologia, Faculdade de Filosofia e Ciências Humanas,
Universidade Federal de Minas Gerais, Belo Horizonte, MG, Brasil
[6]Hospital de Clínicas de Porto Alegre, Porto Alegre, RS, Brasil

*These authors contributed equally to this study.
Corresponding author: A.L.B. Godard
E-mail: brunialt@ufmg.br

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ABSTRACT. Adrenoleukodystrophy is a neurodegenerative X-linked recessive disorder. It is characterized by abnormal function of peroxisomes, which leads to an accumulation of very long-chain fatty acids in plasma and tissues, especially in the cortex of adrenal glands and white matter of the central nervous system, causing demyelinating disease and adrenocortical insufficiency (Addison's disease). It is caused by a mutation in the ABCD1 gene (ATP-binding cassette, subfamily D, member 1), which encodes the

protein adrenoleukodystrophy that is involved in the transport of fatty acids into the peroxisome for degradation. Variable expression has been recognized in families of patients who have this disease. A Brazilian family from Minas Gerais State, Brazil, was studied. The proband is an adult living in Minas Gerais State, Brazil; he had adrenomyeloneuropathy, adrenocortical insufficiency and a stable cerebral form. DNA was extracted from a blood sample and was sequenced to identify the mutation. The patient's exons were cloned for confirmation. A new mutation was found in exon 5 of the ABCD1 gene (c.1430delA), as well as a single-nucleotide polymorphism in exon 6. The mutation causes a frame shift, resulting in a truncated protein with almost total absence of the ATP binding domain.

Key words: Adrenoleukodystrophy; X-linked; ABCD1 gene; Molecular diagnosis; Addison's disease; ABC transporters

