Abstract

X-linked adrenoleukodystrophy (X-ALD) is an inherited peroxisomal disorder caused by mutations in the *ABCD1* gene which codes for an ATP binding cassette transporter. As a consequence of these mutations very long chain fatty acids accumulate in cells and patients develop neuronal and adrenal pathologies. There is a broad phenotypic variability in men suffering from X-ALD but the severity of symptoms is independent of *ABCD1* genotype. Therefore modifier genes and influence of environmental factors were suggested. Although X-ALD was originally referred to as gonosomal recesive, 88 % of heterozygote women over 60 have neurological symptoms. The association of skewed X chromosome inactivation and severity of disease was studied in several publications with conflicting results. Therefore the penetrance and expressivity of X-ALD in women is probably also influenced by other factors. Based on current knowledge future development of the disease cannot be predicted by evaluation of X inactivation patterns.

Key words: X-linked adrenoleukodystrophy, very long chain fatty acids, *ABCD1*, X inactivation, heterozygotes