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X linked recessive ichthyosis: A review

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Abstract

In the present review, we describe the most important aspects of the X-linked ichthyosis (XLI) and make a compilation of the some historic details of the disease. The aim of the present study is an update of the X-linked ichthyosis. Historical, clinical, epidemiological, and molecular aspects are described through the text. Recessive X-linked ichthyosis is a relatively common genodermatosis affecting different ethnic groups. With a high spectrum of the clinical manifestations due to environmental factors, the disease has a genetic heterogeneity that goes from a point mutation to a large deletion involving several genes to produce a contiguous gene syndrome. Most XLI patients harbor complete STS gene deletion and flanked sequences; seven 7 intragenic deletions and 14 point mutations with a complete loss of the steroid sulfatase activity have been reported worldwide. In this study, we review current knowledge about the disease.

Key words: STS gene; X-linked ichthyosis; Steroid sulfatase; Gene deletion; Contiguous gene syndrome; Genodermatosis

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