Abstract

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Journal of Cell and Developmental Biology

2022
Vol 6. No.S4

A novel nonsense mutation in the STS gene in a Pakistani family with X-linked recessive ichthyosis: including a very rare case of two homozygous female patients

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Abstract

Background: X-linked ichthyosis (XLI; OMIM# 308100) is a recessive keratinization disorder characterized by the presence of dark brown, polygonal, adherent scales on different parts of the body surface. It almost exclusively affects males and the estimated prevalence ranges from 1:2000-6000 in males worldwide. Extra cutaneous manifestations are frequent including corneal opacities, cryptorchidism, neuropsychiatric symptoms or others. Up to 90% of XLI cases are caused by recurrent hemizygous microdeletion encompassing entire STS gene on chromosome Xp22.3, while only a minority of patients shows partial deletions or loss of function point mutations in STS. Larger deletions also involving contiguous genes are identified in syndromic patients. Methods & Results: - Here, we report clinical and genetic findings of a large Pakistani family having 16 affected individuals including 2 females with XLI. Molecular karyotyping and direct DNA sequencing of coding region of the STS gene was performed. The clinical manifestations in affected individuals involved generalized dryness and scaling of the skin with polygonal, dark scales of the skin on scalp, trunk, limbs, and neck while sparing face, palms and soles. There were no associated extra-cutaneous features such as short stature, cryptorchidism, photophobia, corneal opacities, male baldness, and behavioral, cognitive, or neurological phenotypes including intellectual disability, autism or attention deficit hyperactivity disorder. Molecular karyotyping was normal and no copy number variation was found. Sanger sequencing identified a novel hemizygous nonsense mutation (c.287G>A; p.W96*), in exon 4 of STS gene in all affected male individuals. In addition, two XLI affected females in the family were found to be homozygous for the identified variant.

Received: April 09, 2022; **Accepted:** April 19, 2022; **Published:** April 27, 2022

Biography

Experienced Researcher with a demonstrated history of working in the higher education industry. On Hands expertise on ABI 3730XL Genetic Analyzer, ABI 7500 Real Time PCR BD LSRII Flow Cytometer, Skilled in Mouse Models, ELISA, Real-Time Polymerase Chain Reaction (qPCR), Genetics, and Cell Biology. Strong research professional with a Master of Philosophy focused in Molecular Biology (Genetics) from Centre of Excellence in Molecular Biology, Lahore, Pakistan

