NOVEL FOXC2 MISSENSE MUTATION IDENTIFIED IN PATIENT WITH LYMPHEDEMA-DISTICHIASIS SYNDROME

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ABSTRACT

Lymphedema-distichiasis (OMIM 153400) is a dominantly inherited disorder typically presenting with lymphedema at puberty and distichiasis at birth. The condition has been decisively linked to mutations in the forkhead transcription factor FOXC2 which have been primarily frameshift mutations truncating the protein. We report here a novel missense mutation along with a literature review summarizing reported mutations.