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A Novel SRY Pathogenic Variant from a 46,XY Female Harboring a Nonsense Point Mutation (G to A) in Position 293

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Abstract

SRY gene mutation is a common cause of 46,XY female. We report a 46,XY female with a novel mutation of SRY c.293G>A (p.Trp98ter). Our report provides evidence for a pathogenic role of the SRY gene c.293G>A mutation in an individual and enlarges the spectrum of molecular diagnosis for these patients.

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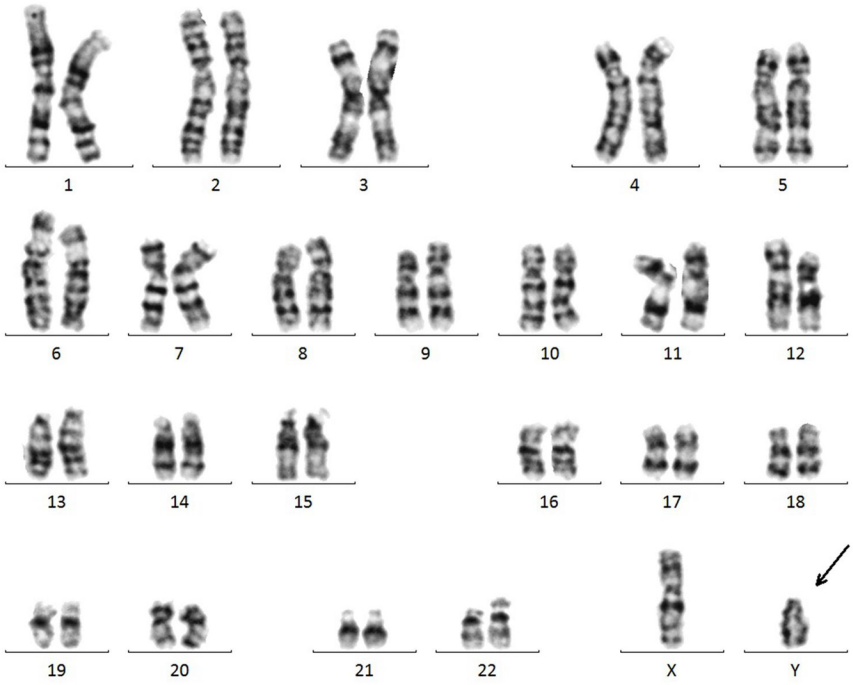

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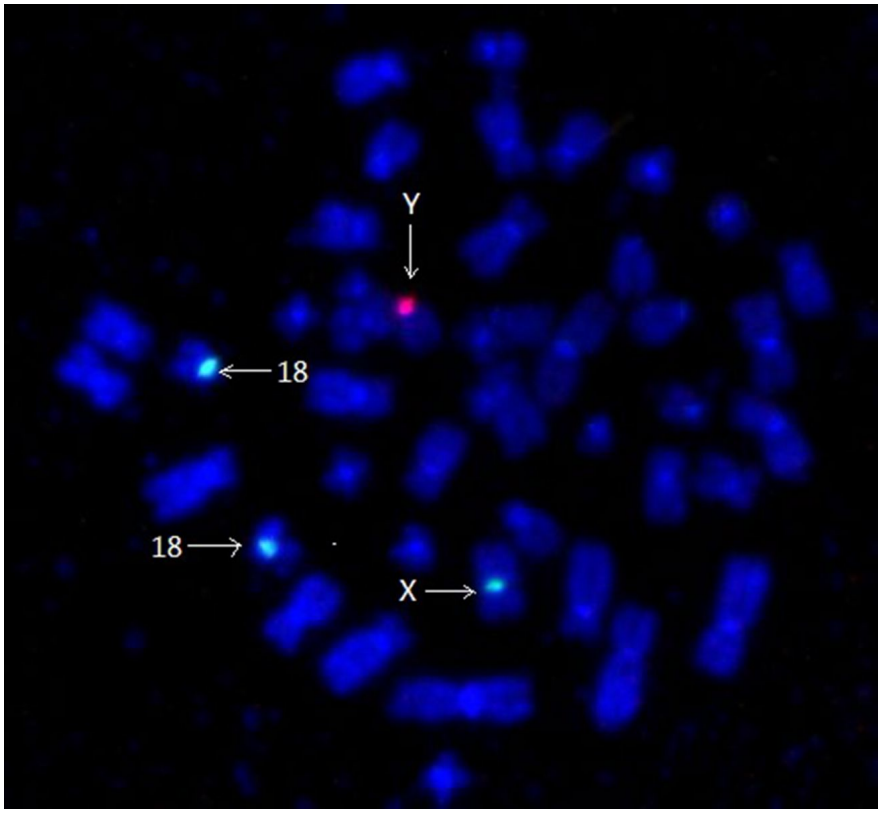

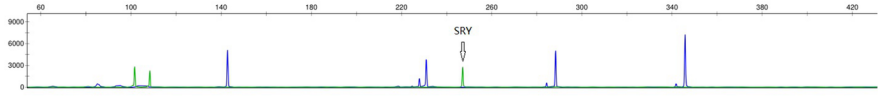

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