

DDX3X Syndrome

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RNA molecules rely on proteins across their life cycle. DDX3X encodes an X-linked DEAD-box RNA helicase with a Y-linked paralog, DDX3Y. DDX3X is central to the RNA life cycle and is implicated in many conditions, including cancer and the neurodevelopmental disorder DDX3X syndrome.

DDX3X-linked conditions often exhibit sex differences, possibly due to differences between expression or function of the X- and Y-linked paralogs DDX3X and DDX3Y. DDX3X-related diseases have different mutational landscapes, indicating different roles of DDX3X. Understanding the role of DDX3X in normal and disease states will inform the understanding of DDX3X in disease. We review the function of DDX3X and DDX3Y, discuss how mutation type and sex bias contribute to human diseases involving DDX3X, and review possible DDX3X-targeting treatments.