

Predicting how splicing errors impact disease risk

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Cells make proteins based on blueprints encoded in our genes. These blueprints are copied into a raw RNA message, which must be edited, or spliced, to form a mature message that can direct the cellular machinery that synthesizes proteins. CSHL scientists have rigorously analyzed how mutations can alter RNA messages at the start of a splicing site (5-prime splice site). 1 and 2 here indicate those positions in a hypothetical raw RNA message. The aim is to be able to predict how errors at these sites will affect protein synthesis. Some errors lead to serious illnesses. Credit: Diagram courtesy of Khan Academy. Note: All Khan Academy content is available for free at (www.khanacademy.org



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