

Mutación en el gen LDB3 - 2018

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La mutación encontrada en el gen LDB3 está asociada a cardiomiopatía hipertrófica, dilatada, no compactada y miopatía miofibrilar y muy probablemente nada tiene a ver con el fenotipo encontrado "overlapping Brugada syndrome/ARVC/D".

Este gen LDB3 que Ramón Brugada ha encontrado tiene por tarea proporciona instrucciones para crear una proteína llamada enlace de dominio LIM 3 (LDB3).

La proteína LDB3 se encuentra en el miocardio y en el músculo esquelético. Las proteínas LDB3 se encuentran dentro del sarcómero (unidad funcional de la contracción y relajación del músculo cardíaco). Esta proteína se une a otras proteínas participando del mantenimiento de la estabilidad de los discos Z que forman el límite de la unidad contráctil. Los discos Z unen a los sarcómeros vecinos para formar miofibrillas de actina (finas) y de miosina (gruesas), interdigitadas forman la unidad básica de las fibras musculares.

La unión de los sarcómeros y la formación de miofibrillas proporcionan la fuerza de contracción y relajación muscular mecanismo conocido como traslapación y destraspación.

Varias versiones diferentes (isoformas) de la proteína LDB3 se producen a partir del gen LDB3. Mutaciones en este gen causan 4 entidades, y la ubicación citogenética se encuentra en el brazo largo del cromosoma 10 (10q23.2) en la posición 23.2.

El gen LDB3 ha recibido varias otras denominaciones: **LDB3Z1, ZASP, LDBEZ4, y LDB3Z4.**

El tema es tan nuevo que hasta la fecha apenas se han publicado 40 artículos en el Pubmed acerca de este gen (Casi todos en 2016, 2017 y 2018. Apenas 1 en 2015):

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