Gen LDB3 y mutaciones

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La mutación encontrada en el gen LDB3 está asociada a cardiomiopatía hipertrófica, dilatada, no compactada y miopatía miofibrilar y que muy probablemente nada tiene a ver con el fenotipo encontrado "overlappying 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 contraccion y relajación del músculo cardiaco). Esta proteína se une a otras proteínas participando del mantenimiento de la estabilidad de los discos Z que forman el limite de la unidad contráctil.
Los discos Z unen a los sarcómeros vecinos para formar miofibrilas 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 translapación y destraslapación. Varias versiones diferentes (isoformas) de la proteína LDB3 se producen a partir del gen LDB3. Mutaciones em este gen causan las 4 entidades mencionadas (cardiomiopatía hipertrófica, dilatada, no compactada y miopatía miofibrilar), y la ubicación citogenética de este gen se encuentra en el brazo largo del cromossoma 10 (10q23.2) en la posición 23.2.
El gen LBD3 há recebido várias otras denominaciones: **LDB3Z1, ZASP, LDBEZ4, y LDB3Z4.**

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

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