Diagnostico prenatal y nueva mutación en enfermedad granulomatosa crónica ligada al cromosoma X.

Pérez-Aradas V., Mancebo E., Talayero P., González-Granado L.I., Allende L.M.
Servicio de Inmunología, Hospital Universitario 12 de Octubre, Madrid, España.

BACKGROUND: Chronic Granulomatous Disease (CGD) is a rare primary immunodeficiency caused by the alteration of the enzyme complex NADPH oxidase, which affects the phagocytic function. CGD patients are susceptible to recurrent infections mainly caused by bacteria and/or fungi.

METHODS: We studied a 6 year-old boy with suspicion of CGD. The diagnosis was confirmed based on the functional study of NADPH oxidase. Simultaneously, the second pregnancy of the mother was reported and genetic counselling was requested.

RESULTS: We identified a new disease-causing mutation by direct sequencing of the CYBB gene (X-linked CGD). The prenatal study resulted in the identification of the same mutation in the foetus.

COMMENTS: Molecular genetics characterisation of CGD is needed to obtain an accurate diagnosis of the disease and to offer prenatal diagnosis and genetic counselling in future pregnancies.

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