Hypertrophic Cardiomyopathy: A New Mutation Illustrates the Need for Family-Centered Care

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This is a case series of a family positive for a previously undescribed mutation in the myofilament gene MYH7, causing hypertrophic cardiomyopathy (HCM), a potentially lethal cardiac disease with strong hereditability. The family's significant disease became strikingly apparent with the unanticipated diagnosis of their newborn infant shortly after her birth. With genetics consultation, this led to the discovery of the MYH7 mutation, in which lysine replaces glutamic acid at amino acid 1752. Further genetic testing revealed the same mutation in her father and two siblings, all of whom had varying degrees of disease severity. Despite prior diagnosis of HCM for the paternal grandmother and great uncles, this family's situation points to the need for continued education of healthcare providers when heritable diseases are encountered. Genetic counseling should occur early and has been shown to be helpful in making an accurate diagnosis and identifying relatives at risk of developing the condition. It may, as in this case series, lead to the discovery of a novel mutation and contribute to the growing genetic database for familial HCM.