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研究报告

22q11微缺失与先天性心脏病的关系的研究

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摘要 应用染色体荧光原位杂交（FISH）技术，对25例不同表型的先天性心脏病患者外周血标本进行22q11微缺失的检测，以探讨先天性心脏病与22q11微缺失的关系。受检的23例单纯性先天性心脏病患者，无22q11缺失者为19例，发生缺失者为4例；2例法鲁氏四联症伴心外多发畸形患者，有22q11缺失。上述结果表明，先天性心脏病与22q11微缺失有关。

关键词 荧光原位杂交；先天性心脏病；染色体微缺失

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Study on the Relationship Between 22q11 Microdeletion and Congenital Heart Disease

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Abstract

To investigate if microdeletion of chromosome 22q11 is an epidemiologically important cause of congenital heart disease (CHD), we studied 25 cases with CHD phenotypes. Venous blood samples were tested by fluorescence in situ hybridization (FISH) for microdeletion of 22q11. Among 23 cases with simple CHD, 19 were shown not to have microdeletion of 22q11 and the other 4 cases were shown to have 22q11 microdeletion. Microdeletion of 22q11 was found in 2 cases with Tetralogy of Fallot (TOF) accompanied by multiple malformations. The results suggested that microdeletion of 22q11 was associated with CHD.

Key words FISH congenital heart disease chromosome microdeletion

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