The Link Between 22q11.2 Deletions and Tetralogy of Fallot

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Tetralogy of Fallot (TOF) is a congenital heart condition that has no known cause, characterized by the following: overriding aorta, ventricular septal defect, right ventricle overflow obstruction, and right ventricle hypertrophy. This condition can be lethal during early life if untreated. Research to better understand the causes of TOF has found a potential connection between TOF and microdeletions on chromosome 22 at location 22q11.2, leading to the question of what role 22q11.2 deletions may have in TOF. These deletions occur in 1 in 4000 people, leading to the mutation being associated with several conditions. To better understand how 22q11.2 deletions may be associated with TOF, this review analyzed their potential role in causing TOF and how they influence treatment outcomes. It was found that 22q11.2 deletions were in high concurrence with TOF. Furthermore, 22q11.2 deletions and TOF were often in co-occurrence with other conditions, including facial abnormalities, learning and mental disabilities, and pulmonary atresia. Lastly, it was found that patients who had both 22q11.2 deletions and TOF had longer hospital stays, more extensive medication and follow-up care, and higher mortality rates. Therefore, this review finds that TOF patients with 22q11.2 deletions will most likely have co-occurring conditions that may cause treatment to be more extensive, yet it is unclear if 22q11.2 deletions are causative of TOF. Therefore, more research is needed to better understand and find other potential genetic causes of TOF by looking at mutations associated with structural defects.