Haploinsufficiency of vascular endothelial growth factor related signaling genes is associated with tetralogy of Fallot

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See the UHN Newsroom article

Studying adults who were born with major heart defects (“blue babies”), but who did not have 22q, we discovered a new pathway to these important conditions. We used the most advanced genetic sequencing methods available and carefully examined for changes to genes that were likely to cause problems with the development of the heart. The findings focussed on a signalling mechanism that may be important for many individuals, including those with 22q.