



Genetic Footprints of Heart Disease



In many cases, the cause of death stems from inherited diseases of the heart's electrical system or the heart muscle. If such cases were diagnosed promptly, it could save many lives and enable clinicians to intervene before a parent or sibling suffers from a cardiac event.

The main problem is that standard autopsies do not reveal inherited cardiac anomalies so it can be fairly difficult to determine whether the disease was inherited or not.

To better improve the detection of inherited cardiac anomalies in families, Gregory Webster, MD, MPH, a cardiologist at Ann & Robert H. Lurie Children's Hospital teamed up with colleagues from Northwestern University Feinberg School of Medicine to trace the footprints of genetic heart disease in young people who died suddenly and whose cause of death had not been determined through traditional autopsy.

The researchers used molecular autopsies to analyse genetic code and search for aberrations linked to cardiac death. While DNA samples are collected by some medical examiners' and coroners' offices, their accuracy remains unclear. The goal of this research was to clarify the accuracy of such tests in the recognition of inherited heart disorders.

By collaborating closely with medical examiners' and coroners' offices, the researchers identified such cases and performed detailed genetic analysis in both the deceased and the survivors. In addition, the survivors also underwent clinical evaluation including a physical exam, electrocardiograms and echocardiograms.

"This study is an opportunity to understand how inherited diseases of the heart affect families," Dr Webster says. "Anytime a child or a young person dies suddenly, it's a tragedy. Findings from this research will illuminate ways to help us prevent other such tragedies from occurring in the same family." He also highlights the fact that identifying family members who can benefit from prompt diagnosis and treatment of inherited heart disease is critical in preventing sudden cardiac death.

Source: Ann & Robert H. Lurie Children's Hospital of Chicago

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