Inherited Cardiac Conditions

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Inherited cardiac conditions are caused by changes in one or more genes passed from generation to generation. They can affect people of all ages or sizes. If left undetected or untreated, an inherited heart condition could lead to heart failure, or sudden death from cardiac arrest. In many cases, the first sign of an inherited cardiac disease is when someone in the family suddenly dies with no obvious cause or explanation. According to the Centers for Disease Control and Prevention, about 735,000 Americans have a heart attack every year (with about 610,000 dying from it). Over the years, the numbers continue to increase. As our society continues to grow and develop, we can continue to solve our problems using STEM-based solutions. For example, in order to prevent inherited cardiac conditions in the future, scientists and engineers having been working for the past decades to develop technologies that can trace genetic mutations in cells early on, and eradicate it.

To begin with, over the years, scientists and doctors have uncovered the science behind inherited cardiac conditions. It is traced back to genetics. Genes are the basis of inheritance, and are composed of deoxyribonucleic acid (DNA). They are in charge of many functions, including providing instructions to make proteins. Humans have over 20,000 genes, and each and every gene is present in two copies. One copy being inherited from the mother, and the other inherited from the father. Because genetic conditions are caused by a mutation in one or more genes passed from generation to generation, most genetic heart conditions are inherited in an autosomal

dominant pattern. An autosomal dominant pattern is a pattern of inheritance in which an individual, who is affected, receives one copy of a mutant gene and one normal gene on a pair of autosomal chromosomes.

Furthermore, scientists and doctors have discovered a heterogeneous spectrum of cardiac diseases, including cardio-myopathies and arrhythmic diseases occurring in structurally normal hearts. According to Hagerstown Cardiovascular Medicine, “The extraordinary progress achieved in molecular genetics over the last three decades has unveiled the complex molecular basis of many familial cardiac conditions, paving the way for routine use of gene testing in clinical practice. In current practice, genetic testing can be used in a clinically affected patient to confirm diagnosis, or to formulate a differential diagnosis among overlapping phenotypes or between hereditary and acquired (non-genetic) forms of disease. Although genotype–phenotype correlations are generally unpredictable, a precise molecular diagnosis can help predict prognosis in specific patient subsets and may guide management (*Girolami*).” Over the years, scientists have developed varying genetic tests. As of now, they can clarify the diagnosis in a person, trace the cause of heart disease in a family, and predict which family members are at-risk of carrying the family's heart condition. They can even provide options for family planning, including preimplantation genetic diagnosis, which can allow one to avoid passing a disease-causing mutation to offspring. To be more specific, genetic testing is the process of taking a sample of DNA to look for changes that could cause inherited heart disease. Significant changes in genes are called pathogenic mutations. “Pathogenic” meaning “disease-causing”. For example, in a procedure called a buccal smear, a small brush or cotton swab is used to collect a sample of cells from the inside surface of the cheek. The sample is then sent to a laboratory where technicians look to find important changes in chromosomes, DNA, or proteins, depending on the suspected

disorder, in this case, inherited cardiac conditions. The laboratory will then report the test results in writing to a person's doctor or genetic counselor, or directly to the patient if requested.

Moreover, in efforts of eliminating cardiac conditions early on, scientists and doctors have developed preimplantation genetic diagnosis (PGD). Preimplantation genetic diagnosis is a procedure used prior to implantation in order to identify any genetical defects with embryos. This process prevents genetic diseases from passing on to offsprings. According to the American Pregnancy Association, " Preimplantation genetic diagnosis involves the following steps: First, a couple/few cells are micro-surgically removed from the embryos, which are about 5 days developed. After this cell collection, the embryos are safely frozen. The DNA of the cells is then evaluated to determine if the inheritance of a problematic gene is present in each embryo. This process takes at least one full week. Once PGD has identified embryos free of genetic problems, the embryo(s) will be placed in the uterus (usually by an IVF procedure), and the wait for implantation and a positive pregnancy test begins. Any additional embryos that are free of genetic problems are kept frozen for possible later use while embryos with the problematic gene(s) are destroyed. This testing process may take weeks (*American Pregnancy Association*)." Preimplantation genetic diagnosis can benefit possible carriers of a disease at risk of passing it on.

Personally, inherited cardiac conditions run in my father's side. When my father was young (around the age of 11), his grandfather had died from a heart attack. Due to his old age, and high cholesterol, nobody suspected his sudden death. (In older people, having high cholesterol is common for aging causes your heart and blood vessels to stiffen.) Many years later, when my father was in his 30's, his father had also died from heart attack. This sudden death surprised his family. His father was a perfectly healthy man, 50 years of age. After this tragedy, my father

along with his mother and sister, went to get tested. The results came back negative, so they were safe. However, years later, my father's uncle who hadn't gotten tested had died from a cardiac arrest. It had turned out that both my father's father and uncle had both carried the disease from their father. Had his uncle gotten tested, they might have been able to save his life.

To conclude, inherited cardiac diseases are life threatening; killing an estimated 610,000 people per year in the U.S. alone. By tracing it back to its origins, doctors have been able to find the cause of inherited heart conditions. In efforts of preventing inherited cardiac diseases in the future, scientist and engineers have worked together to create the technology needed to find mutations. Genetic tests, preimplantation genetic diagnosis (PMG), and many other procedures, have been discovered using STEM. Although back then, genetic tests were not widely known of, they have become far more accurate. By educating others about genetic disorders, and how they spread, we can ultimately eliminate them.

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