Premature cardiovascular deaths

To the Editor: Sudden death often, but not always, affects young and ostensibly healthy people. The effect on families and friends may be devastating. It is imperative that the cause of the sudden death is determined because other members of the family may be affected with the same heritable condition. We were pleased to read about the important work done on genetic testing for arrhythmogenic disorders,[1,2] and wish to broaden the consideration of causal conditions to several metabolic conditions that should be recognised on clinical and biochemical grounds, especially familial hypercholesterolaemia (FH). FH, despite its high prevalence in South Africa due to founder effects, is often not diagnosed, and treatment is often started late in life, with many patients not achieving their low-density lipoprotein (LDL) cholesterol target.[3]

FH is not rare; a global call for action was made to identify people with this heritable condition as, in most cases, it can be treated with safe and affordable medication.[4] The average age of a first cardiovascular event in subjects with FH was 45 years in a local study, but events may occur at an even younger age.[5] Anecdotally, sudden death occurred as a result of an acute myocardial infarction in a 23-year-old male who had not previously been diagnosed with (heterozygous) FH, despite a family history of premature heart disease. Women with heterozygous FH may also die prematurely: a 26-year-old woman died as a result of an acute myocardial infarction within hours of the delivery of her first child, and a 28-year-old woman with a short history of angina pectoris also died suddenly following a myocardial infarction. Homozygous FH is associated with cardiovascular deaths at an even younger age, but the prognosis has improved with modern treatment.[6] Other lipoprotein disorders that may present like homozygous FH include phytosterolaemia, autosomal recessive hypercholesterolaemia, dysbetalipoproteinaemia[7] and very low concentrations of high-density lipoprotein (HDL) cholesterol.[8]

There are also connective tissue disorders affecting the arterial system that may result in rupture and/or thrombosis with haemorrhage, dissection, or coronary or cerebral arterial events.[9] Toxins may also play a role, including cannabis, cocaine and even psychotropic drugs.[10] In the context of a sudden (cardiac) death in athletes, there are several additional considerations,[11] including commotio cordis.[12]

We hope that a sudden, unexpected death will prompt a thorough clinical, biochemical, pathological and genetic investigation to identify its cause so as to prevent recurrences of the same tragedy in other family members, by preventive treatment, preparation for resuscitation and to provide closure for the family of the deceased. FH is an important treatable and preventable cause of sudden death.

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