Yield from family screening in a national adolescent cardiac screening program

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Introduction: Cascade family screening in patients with confirmed or suspected inherited cardiac disorders is now well established. This may refute or confirm a familial clinical diagnosis and is particularly relevant in young adolescent individuals as it may be too early to manifest a distinct phenotype.

Objectives: A large cohort of 2708 adolescents aged 14–16 years gave consent to participate in a national cardiac screening program (BEAT-IT). Individuals with suspected inherited cardiac disorders were extensively evaluated. Their relatives were also invited to undergo screening. This study reports the yield of this family cardiac screening program.

Methodology: Family members of probands with suspected or confirmed inherited cardiac conditions were offered cardiac screening. A standard clinical screening protocol for all first-degree family members included a resting 12-lead ECG and echocardiogram. Those with a channelopathy suspicion also underwent postural ECGs and exercise testing. Screening second-degree relatives was also performed in a cascade fashion when clinically indicated. Relatives with a normal baseline screen were offered surveillance if younger than 25 years or a proband clinical diagnosis. Those with an abnormal ECG and/or echocardiogram were referred for further evaluation.

Results: 17 probands (63% females) were suspected of harbouring inherited heart disease. Another 2 were diagnosed with a clinical phenotype. The mean age was 15.3±0.58 years. All were Caucasian.

77 family members underwent cardiac screening, with a mean age of 42.5±16.43 at first evaluation. The majority were female (n=44, 57.1%). 12 (15.6%) had an abnormal ECG. 6 (7.8%) had an abnormal echocardiogram, with 2 (2.6%) consistent with cardiomyopathy. 8 (10.4%) were diagnosed with an inherited cardiac condition (n=2 HCM, n=1 DCM, n=5 LQTS). Another 7 (9.1%) are under surveillance because of a pathological ECG in the absence of a clinical phenotype.

The highest clinical yield was in the Long QT group (n=5, 55.6%). Family members (n=25) referred because of proband lateral TWI were the second most likely to require clinical follow-up because of a pathological ECG or a clinical diagnosis (n=7, 28.0%). Relatives referred because of isolated anterior TWI on the proband's ECG had the lowest diagnostic yield (n=17, 0%). After excluding families of probands with isolated anterior TWI (n=18), the overall clinical yield increased to 13.6%. Another 11.9% are under surveillance because of a pathological ECG.

Conclusion: The yield of family screening as part of a national cardiac screening program was 10.4%. This increases to 13.6% when excluding probands with anterior TWI, with 11.9% under surveillance because of a pathological ECG. To our knowledge, this is the first such study of its kind.