IDENTIFYING INDIVIDUALS AT RISK FOR ARVC CAUSED BY TMEM43 P.S358L: A GENETICS EDUCATIONAL TOOL FOR PRIMARY CARE PHYSICIANS

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ABSTRACT: Poster (P-21)

Background: Genetics and genomics medicine is becoming integrated into primary care. While many Primary Care Physicians (PCPs) report a lack of confidence in referring and counselling patients, knowledge, experience, and skills to adequately fulfill their role, PCPs acknowledge this integration. Newfoundland and Labrador (NL) has a high incidence of arrhythmogenic right ventricular cardiomyopathy (ARVC) caused by the *pS358L TMEM43* mutation. Its first indication can be sudden cardiac death (SCD). Timely referral of at-risk individuals to genetic/cardiac services is critical as correct diagnosis significantly reduces mortality by enabling effective treatment with an implantable cardioverter defibrillator. Short, concise summaries of genetic conditions that highlight "red flags" are effective tools for educating PCPs. However, no tool currently exists for this subtype of ARVC.

Methods:

1. Draft tool reviewed with a physician focus group
2. Based on feedback, family history data from 26 known families in NL analysed to inform next iteration
3. Revised tool will be distributed to PCPs and Family Medicine Residents in NL for review before final tool modifications and distribution

Results: A physician focus group identified that the tool needed to be concise and provide clear referral warning signs. The 26 known families were assessed for the number of relatives (with SCD ≤50yrs) of affected individuals (n=409). Of these 409, 160 were diagnosed following SCD (i.e., unrecognised as being at-risk prior to their final symptom), of which 56%, 39% and 31% had at least one first, one second and one third degree relative presenting with an SCD (≤50yrs) respectively, with 19% having no known affected relatives. Family history of young SCD therefore is a significant 'red flag' and will inform the tool's next iteration.

Conclusions: Extended family histories provided quantitative concise information to update the tool and provide one clear referral guideline for immediate referral to specialists.