Investigation of current models of care for genetic heart disease in Australia: A national clinical audit


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Background

Genetic heart diseases are a major cause of morbidity and account for over half of all sudden deaths in young people. They are often heterogeneous and clinical management is complex. Best clinical practise is achieved with a multidisciplinary team approach.

The Australian Genomics Cardiovascular Genetic Disorders Flagship broadly aims to demonstrate the impact of whole genome sequencing in clinical care. This audit is a sub-study of the flagship and conducted the first nation-wide patient record audit in Australia to establish current practices across cardiac genetics clinics.

Method

A random selection of patient records were audited by trained researchers if they met the following criteria:

- suspected cardiomyopathy, primary arrhythmia, or autosomal dominant congenital heart disease (CHD) and;
- had a public cardiac genetics consultation between 1st January 2016 and 31st July 2018 and;
- were offered a diagnostic genetic test.

Ten percent of audits underwent Inter-rater reliability (IRR). There was a good concordance amongst auditors (kappa coefficient >0.6).

Results

Demographics: There were 536 patient records (electronic and hardcopy) audited from eleven public tertiary hospitals across five states (Figure 1). The mean age of patients was 45 years old and 53.6% were male. Nationally, most patient lived in metropolitan areas (64.8%). Queensland had the largest number of patients from regional and remote areas (44.2%), followed by Victoria (41.6%) (χ² 66.5, P<0.001).

Clinical diagnosis: Cardiomyopathies accounted for 65.7% of all diagnoses (Figure 2). Other diagnoses were cardiac arrest, conduction disease and unclassified. The most common reason for a diagnosis was the investigation of symptoms (75.4%), followed by family screening (13.8%) and incidentally (e.g. a work medical) (10.6%).

Referrals: In total, 85.3% were from Cardiologists, followed by GPs (9.0%).

Mode of consultation: Across all sites, outpatient clinics (90%) were the most frequent; 6% were inpatient consultations and 4% telehealth. Queensland had the highest proportion of telehealth (9%).

Genetic testing: targeted panels were the most requested test (70%); whole genome sequencing accounted for 7%. Most tests were funded by the genetics clinic (72.8%) and several national and international testing laboratories were utilised. i.e. Path West (31.6% of all tests).

In total, 30% of tests identified a variant that explained the diagnosis. Figure 3 shows the detection rate for each diagnosis (the difference was not statistically significant). The most frequently affected gene in the cardiomyopathies was MYBPC3 and in the Arrhythmias it was KCNH2 and KCNQ1.

Figure 3: Proportion (%) of Pathogenic variants and variants of uncertain significance (VUS) found for each clinical diagnosis category

Conclusion

We provide important information describing the current models of care for genetic heart diseases throughout Australia. The baseline data will inform the implementation and impact of whole genome sequencing in the Australian genetic heart disease healthcare landscape.