Novel Development of an Inherited Cardiac Conditions Service in a District General Hospital

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Introduction

Genetic causes of inherited cardiac disease (ICC) with significant implications for patients and families are increasingly recognised. Historically, genetic testing has been expensive, time consuming and mainly confined to London-based tertiary referral centres [(1)].
However, new advances in DNA sequencing and large volume laboratories have made testing more accessible. Indeed, recent government policy places genomic, personalised medicine at the forefront of routine healthcare to improve the prevention, diagnosis, stratification and treatment of disease [(2)].
There is therefore a nationwide move towards 'mainstreaming' of genetic testing. To support this expansion, the National Genomic Test Directory [(3)] describes which patients should undergo genetic screening and the appropriate genetic panel for each phenotype.

In parallel, the technology to deliver remote MDT working and data sharing has advanced significantly. The recent 'Getting It Right First Time' report in cardiology [(4)] suggests a regional network model for the heart team – including the default use of virtual MDTs with access to appropriate data and expertise to facilitate decision making. This would improve equitable, timely access to services regardless of geography.
The economic and environmental costs of long-distance patient travel are also falling under increasing scrutiny.

We describe the establishing of a new programme for genetic testing for ICCs within a small general hospital cardiology department.

Demographics

N	50
Age at referral (years)	55 ±22
Sex - male	38 (76%)
HCM	10 (20%)
DCM	26 (52%)
ARVC	1 (2%)
Other	11 (22%)
Cascade screening	2 (4%)
First degree relative affected	10 (20%)
NYHA III/IV	7 (14%)
NT Pro BNP (ng/L)	1909 ±4351
Positive genetic result	8 (16%)
Ventricular arrythmia	12 (24%)
Discussed in regional MDT	9 (18%)
ICD recommended	7 (14%)
Died or transplant	3 (6%)

Figure 1 - Cohort Demographics

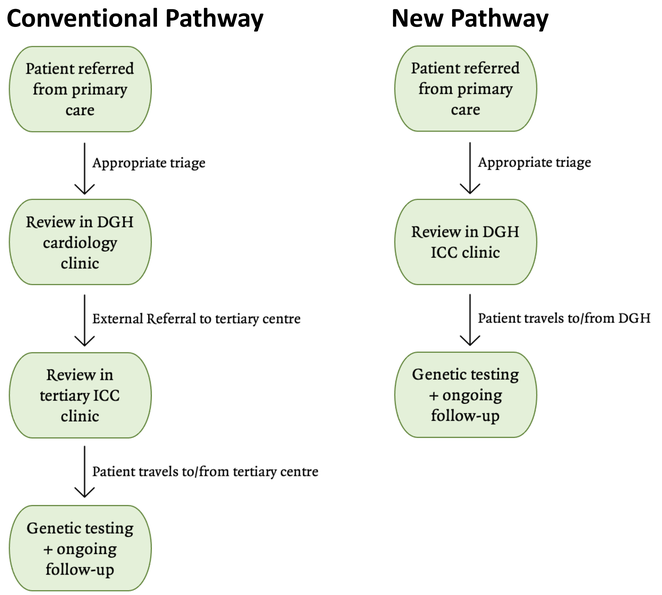

Figure 2 - The new, local inherited cardiac conditions service framework. If patients are appropriately triaged, they can be referred direct from primary care. They can then have local genetic screening and ongoing follow-up

Methods

The service was established in March 2020. We reviewed all patients undergoing genetic testing to October 2021. The time, cost and carbon emission savings for patients accessing this service were compared to conventional referral and follow-up at the nearest tertiary centre. Distances and travel time were calculated via car using Google Maps. Fuel costs were calculated based on an average of 30 miles per gallon and £1.40 per litre of fuel.
Carbon emissions were based on a car with this economy driving the calculated distances using an online carbon footprint calculator (https://www.fleetnews.co.uk/costs/carbon-footprint-calculator/).
Non-parametric data is presented as median (±IQR). Parametric data is presented as mean (±SD).
When comparing medians of non-parametric data, significance testing was undertaken using the Mann and Whitney U test. All p values of <0.05 were considered statistically significant. Significant results are denoted by an asterisk.

Results

Fifty patients underwent genetic analysis. Median age at referral was 55 (IQR 22). 38 (76%) were male. The mean wait for an appointment was 10.5 weeks (±6.1). 9 patients (18%) were discussed in a regional MDT. The mean wait for genetic panel results was 3.3 months (±6.1). 10 patients (20%) had HCM. 26 patients (52%) had DCM. 1 patient (2%) had ARVC. 11 patients (22%) had other diagnoses such as arrythmia syndromes or amyloidosis. 2 patients (4.4%) had phenotypically normal hearts and were undergoing genetic cascade screening. 8 patients (16%) returned a positive result - 7 pathogenic or likely pathogenic and 1 variant of uncertain significance. 7 patients (14%) were referred for an ICD. 3 patients (6%) died. The mean number of appointments was 3.7 (±2.6).

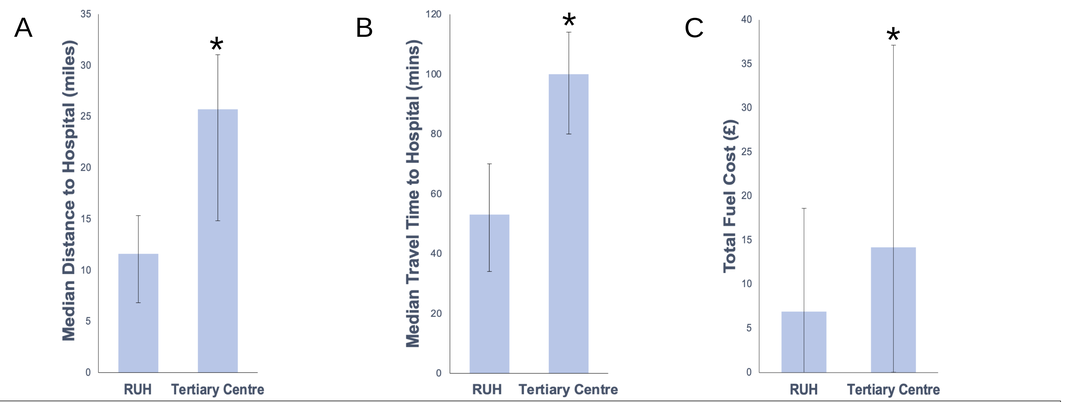

Figure 3 - Practical considerations for the patient - Comparing attending the RUH to the nearest tertiary centre. **A+B -** Distance and return travel times would be significantly longer to attend the tertiary centre - presented as the median (±IQR). **C-** Total fuel cost per patient undergoing follow-up after the initial appointment presented as median (±IQR). This is a vital consideration in providing equitable, accessible care with significantly rising fuel costs.

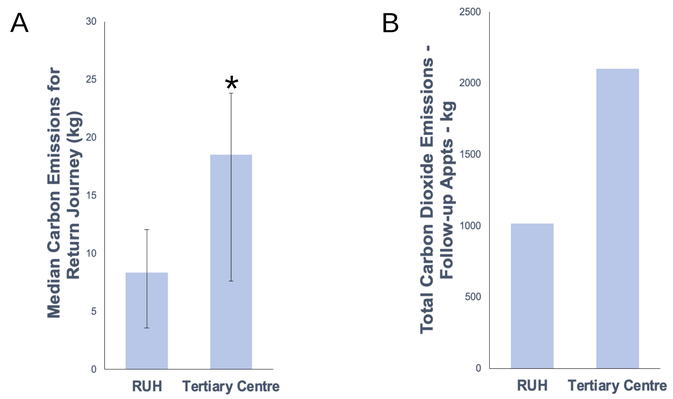

Figure 4 - Carbon emissions. A- Median carbon emissions per each return journey to the RUH or the nearest tertiary centre presented as the median (±IQR). This more than doubled if patients had to travel to the tertiary centre. This will compound with repeated visits for follow-up or surveillance.
B- Total theoretical carbon savings presented for all patients undergoing follow-up - a projected 1,085kg saving in this small cohort.

Conclusions

The advent of precision medicine, wider recognition of ICCs and improved availability of genomic sequencing will generate a significant clinical need. Care networks will therefore need to be developed to ensure equitable access to services. It is cost effective, sustainable, and convenient for patients to be cared for at their local hospital. The service can be facilitated by remote MDT if tertiary opinion is required. This service is dependent upon local ICC expertise.

References

1. Burton H, Alberg C, Stewart A. Mainstreaming genetics: a comparative review of clinical services for inherited cardiovascular conditions in the UK. Public Health Genomics. 2010;13(4):235-45. doi: 10.1159/000279625. Epub 2010 Apr 15. PMID: 20395692. **2.** UK Government Policy Paper - Genome UK: The Future of Healthcare. 2020. https://www.gov.uk/government/publications/genome-uk-the-future-of-healthcare/genome-uk-the-future-of-healthcare. Viewed 24/11/21. **3.** NHS National Genomic Test Directory Testing Criteria for Rare and Inherited Disease. 2020;(August):280. Available from: https://www.england.nhs.uk/publication/national-genomic-test-directories/%0Ahttps://www.england.nhs.uk/publication/national-genomic-test-directories/%0Ahttps://www.england.nhs.uk/wp-content/uploads/2018/08/Rare_and_Inherited_Disease_Eligibility_Criteria_Aug. **4.** Cardiology Getting It Right First Time Programme National Specialty Report. Clarke S, Ray S. February 2021. https://www.gettingitrightfirsttime.co.uk/wp-content/uploads/2021/08/Cardiology-Jul21k-NEW.pdf . Viewed 24/11/21.