Introduction

The genetics genealogy team provide a unique service, supplying accurate and comprehensive family history research for patients who have been referred to a clinical genetics service due to a possible risk of familial cancer or other inherited genetic conditions. The determination and management of risk is dependent on the quality of the family information genetics clinicians have to work from. Risk assessment for patients may result in gene testing, prophylactic surgery, screening appropriate to the determined risk or reassurance and discharge from the care of clinical genetics services. For the NHS, determining more accurate genetic risk and managing it appropriately, means limited and expensive resources are used in the most effective manner.

Background

Cancer registries throughout the United Kingdom have, for many years, provided information to genetics clinics on cancer incidence in deceased individuals or, on receipt of consent, for living patients. In terms of this report such requests will be referred to as ‘ad hocs’. What makes the Scottish genetics genealogy service unique however is the research of Scotland’s family history records of births, deaths and marriages, old parish records and census returns to create accurate family pedigrees. Where appropriate we add cancer registration data from the Scottish Online Cancer Registration and Tumour Enumeration System (SOCRATES), giving tumour incidence date(s) and more detailed diagnosis information.
The genealogical research of the family histories is made possible in Scotland due to:

**Availability** – All Scottish statutory records can be accessed after the payment of a fee. In England and Wales, although the indexes are free, sight of the official records are by purchasing them individually.

**Accessibility** - Scottish records are accessed using a powerful computerised search engine and digitised imaging, which we can access via a remote web link to National Records Scotland (NRS).

**Quality** - Scottish records are more informative than in England in that they include maiden names and dates and places of marriage.

The genetics clinics see referrals for a wide range of conditions. The largest single group is cancer followed by cardiac conditions, but neuromuscular disease and learning difficulties also make up a significant proportion of referrals (see figure 1). The clinics also see patients from families with a diverse range of other genetic conditions including hypercholesterolemia, neurological disease and endocrine disorders.

![Figure 1: Referral by Condition to Scottish Genetics Clinics](image)

Although cancer genetics accounts for the vast majority of our work, we also receive requests for different conditions (see figure 2). The number of cardiac requests has stayed broadly the same as last year. The number of neuromuscular requests accounts for a small percentage but this has decreased compared to last year. Neuromuscular requests tend to come in batches when information held by clinics on this patient group is being reviewed so it’s likely the clinics haven’t had any large reviews this year.

The requests in the ‘other’ category increased and are for a variety of conditions, namely Huntington’s disease, Parkinson’s disease, aneurysms, dementia, hypercholesterolemia, brain haemorrhage, thrombosis, malignant hyperthermia and Alpha-1 antitrypsin deficiency.
The non-cancer requests we receive are for those where the conditions are likely to be recorded as a cause or contributing factor of death. We do not have enhanced information from an NSS database for non-cancer requests as we do with SOCRATES for the cancer requests. However providing validated pedigrees with causes of death is still extremely useful for clinicians when trying to assess a patient’s risk of disease and decide on appropriate screening or treatment.

![Figure 2: Referral by Condition to NSS 2018](image)

### Challenges

The primary challenge faced in 2018 was a change in staffing due to a team member retiring. This resulted in a period of time at the end of the year with a vacant post and consequently had an impact on the number of requests we were able to complete. The vacancy was advertised and we look forward to welcoming the successful candidate to commence training with the genetics genealogy team in January 2019.

At the beginning of the year a new NRS system was launched. This appears to be more stable than the previous system. However time was required to familiarise ourselves with the system changes, along with time spent testing and identifying initial problems and errors.

### Ad Hoc Requests

The majority of referrals we receive are from Scotland but we also receive ‘Out of Scotland’ requests from the rest of the United Kingdom, Ireland and occasionally as far afield as Australia and New Zealand. Sometimes clinics may need information on a few individuals within a family and rather than request a full pedigree search they will ask for the information they have to be matched against SOCRATES. The returned information may
be sufficient for them to make a meaningful risk assessment. In many cases however, the returned information may initiate the clinician to request a full genealogical family history search. Alternatively we often find that an ad hoc request cross references to a previously researched family tree and in these circumstances the clinic will receive this additional information, which can assist further with risk assessment.

In accordance with most cancer registries in the UK the number of matches that cannot be made on the initial search of SOCRATES is about 30%. This is due to:

1. Inaccurate and/or incomplete information provided by the patient
2. Possible cancer incidence before reliable cancer registration
3. Misinformation on cancer diagnosis

Unlike other United Kingdom cancer registries we follow up these ‘no traces’ by using statutory records to improve the quality of the information before sending death certification information or trying for a second match with SOCRATES. This results in a remarkably low no trace level which was just over 3% in 2018. The clinics will either receive death certification information and SOCRATES matching or death certification information alone.

Figure 3 shows that in 2018 we received 1,307 Ad Hoc requests and researched 1,257, a slight decrease on the numbers received and researched in 2017. This is likely to be due to the fact that we received very few neuromuscular requests this year.

Figure 3: Ad Hoc Requests Received and Researched

![Figure 3: Ad Hoc Requests Received and Researched](image)
Pedigree Research

The ISD genetics service offers a unique Scottish genealogical research facility that can be used to create and validate family pedigrees, incorporating information and generations beyond the immediate knowledge of the informant. Combined with access to Scottish Cancer Registration data this is an extremely powerful research tool that can have a significant impact on the risk group a patient is assigned to with regards to possible future development of a familial cancer. In 2004 it was established that the patient’s risk category changed in over 29% of cases after pedigree research (Brewster DH et al. Impact of a cancer registry-based genealogy service to support clinical genetics services. Familial Cancer 3: 139-141, 2004). This shows the importance of family validation, not only for clinicians applying appropriate management strategies, but also by reducing healthcare costs by eliminating screening procedures that are not required and may be invasive (Reis MM et al. Analysis of referrals to a multi-disciplinary breast cancer genetics clinic: practical and economic considerations. Familial Cancer 5: 297-303, 2006).

By its very nature such research is labour intensive. It is also difficult to calculate how much time it takes to complete a family as the depth of research is dependent on what is found. This creates challenges as to how much work we can complete with a finite staffing level.

Figure 4 shows that in 2018 we received a similar number of pedigrees to last year but researched less. This was likely due to decreased staffing levels towards the end of the year and resulted in an increase to the waiting list from 96 to 133 family pedigree requests.

The trend identified in previous years of clinics favouring ad hocs over pedigrees has continued. The turnaround for ad hocs is rarely more than two weeks, but pedigrees can take several months, so clinicians may be favouring ad hocs if possible due to the speedier response. Ad hocs requests often contain requests for several individuals so can still involve a significant amount of research and are often subsequently resubmitted as a pedigree request depending on initial findings.

Figure 4: Family Pedigree Requests Received and Researched

![Figure 4: Family Pedigree Requests Received and Researched](chart_image)
Conclusions

2018 has been a good year for the genetics genealogy team. We have ensured any urgent requests are returned in a timely manner to allow critical patient management decisions to be made. We've researched a greater number of non-cancer requests compared to previous years and the ad hoc no trace level remains very low.

The Scottish genetics genealogy service continues to be an extremely valuable resource, which genetics clinics have used to make the most informative choices in the risk management of their patients.

We are grateful to National Services Division for funding the service, the Scottish Cancer Registry for providing such accurate data and National Records Scotland for their comprehensive provision of family records.

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Further Information

Further information can be found on the ISD website

About ISD

Scotland has some of the best health service data in the world combining high quality, consistency, national coverage and the ability to link data to allow patient based analysis and follow up. Information Services Division (ISD) is a business operating unit of NHS National Services Scotland - and has been in existence for over 40 years. We are an essential support service to NHSScotland and the Scottish Government Health Department and others, responsive to the needs of NHSScotland as the delivery of health and social care evolves.