Title

A prospective cohort study assessing clinical referral management & workforce allocation within a UK regional medical genetics service.

Running title - Referral management and workforce allocation.

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Abstract

Ensuring patient access to genomic information in the face of increasing demand requires clinicians to develop innovative ways of working. This paper presents the first empirical prospective observational cohort study of UK multi-disciplinary genetic service delivery. It describes and explores collaborative working practices including the utilisation and role of clinical geneticists and non-medical genetic counsellors. Six hundred and fifty new patients referred to a regional genetics service were tracked through 850 clinical contacts until discharge. Referral decisions regarding allocation of lead health professional assigned to the case were monitored, including the use of initial clinical contact guidelines. Significant differences were found in the cases led by genetic counsellors and those led by clinical geneticists. Around a sixth, 16.8% (109/650) of referrals were dealt with by a letter back to the referrer or re-directed to another service provider and 14.8% (80/541) of the remaining patients chose not to schedule an appointment. Of the remaining 461 patients, genetic counsellors were allocated as lead health professional for 46.2% (213/461). A further 61 patients did not attend. Of those who did, 86% (345/400) were discharged after one or two appointments. Genetic counsellors contributed to 95% (784/825) of total patient contacts. They provided 93.7% (395/432) of initial contacts and 26.8% (106/395) of patients were discharged at that point. The information from this study informed a planned service re-design. More research is needed to assess the effectiveness and efficiency of different models of collaborative multi-disciplinary working within genetics services.

Keywords (MeSH terms)

Genetic Services, Genetic Counseling, Interdisciplinary Communication, Cohort Studies, Delivery of Healthcare, Referral and Consultation.

Introduction

Over the last few years advances in technology have resulted in a rapid increase in the number of gene tests available. In parallel there has been an increase in the number of referrals to genetic services due to heightened public and health professional awareness of the potential of genomic information[1,2]. Recent work indicates that workforce interventions within service re-organisation; including telephone counselling by genetic counsellors [3], the provision of more genetic specialists in rural areas [4]and changes to the referral management systems [5] can have a significant effect on the ability of patients to access genetic service[6,7]. A systematic literature review, entitled "Interventions to improve patient access, service utilization and cost of providing genetic counselling services" is currently planned [7].

The present study describes the usual care pathways of a UK regional genetics service in 2011-2012. One study aim was to monitor the use of recently introduced new initial clinical contact guidelines (ICCG) with the aim of reducing the proportion of home visits and increasing the proportion of telephone contacts. The service provided 26% of its initial contacts by a home visit in 2010-2011.

Within UK regional genetic services clinical patient contact is provided by a multidisciplinary team "a team of professionals including representatives of different disciplines who coordinate the contributions of each profession, which are not considered to overlap, in order to improve patient care" [8]. Disciplines represented Include medical professionals (clinical geneticists), genetic counsellor professionals (genetic counsellors) and nursing professionals (registered nurses) [9].

A systematic review of research literature investigating the role of the non-medical genetic counsellor indicated that genetic counsellors, "undertake a significant workload associated with direct patient care and this appears to be acceptable to patients" [10]. The proportion of patients seen by genetic counsellors without a medical practitioner being present ranged between 39% and 50% and included patients with up to 79 different conditions. However no empirical research studies of the genetic counsellor role were found from any of the European countries. Therefore one aim of this research was to investigate the proportion of patients allocated to each discipline and provide evidence of multidisciplinary working practices. Optimisation of workforce skill-mix could increase the capacity of genetic services, enabling improved access for patients.

Currently the utilization of non-medical genetic counsellors varies across Europe and many countries only have a small number of practitioners[11]. In order to safeguard the public and standardise practice, the European Board of Medical Genetics has defined a code of practice and registration system for genetic counsellors across Europe[12]. English regional genetic services receive centralised funding[13]. The services are provided free to patients at the point of access. The NHS England Medical Genetics Clinical Reference Group has responsibility for defining English service specifications and outcomes for the 23 regional genetics services[14]. UK genetic counsellor professional registration is regulated by the UK and Eire Genetic Counsellor Registration Board. It is based on the principle that in addition to prior academic and vocational qualifications, registered genetic counsellors are required to have a 2-year training period and a Masters level portfolio assessment to become appropriately competent[15]. In 2011 there were 272 genetic counsellor positions in the UK, but not all practitioners were registered with the GCRB [16]. The current number of practitioners is

unknown but 186 are UK Genetic Counsellor Board Registered. The majority working as part of a multi-disciplinary team attached to regional genetics services[17].

The situation across Europe is complicated by differing economic policy, healthcare professional roles, cultural preferences and political systems. Patient surveys show that there is great variability in access to genetic healthcare for diagnosis and on-going treatment [18;19]. To meet this demand clinicians are moving away from established models of service delivery and are developing innovative ways of working such as the integration of genetic and genomic testing within mainstream clinical specialty areas[20]. This refinement of skill mix and workforce optimisation has been seen within other areas of healthcare typified by rising consumer demand and limited resources[21-23]. Historically the UK non-medical genetic service workforce provided psychosocial support and counselling which originated in many services from a community family nursing perspective. This aimed to provide on-going holistic care to families affected by genetic conditions. This focus on continuity of care in a community setting has meant that some services still retain the option of offering a home visit in certain circumstances if indicated by local protocols. This study was designed to describe practice within a regional genetics service and aimed to:

- Determine the proportion of patients allocated to clinical geneticist or genetic counsellor led care.
- Assess the usefulness of initial clinical contact guidelines (ICCG) to aid allocation of telephone, clinic or home visit for the initial contact.
- Describe the range of clinical contacts within a regional genetics service.

- Explore whether a genetic patient reported outcome measures questionnaire[24] and a patient satisfaction questionnaire[25] can be incorporated into service provision.

Methods

This observational prospective cohort study was undertaken in a UK regional genetics service serving a population of 2.7 million, with 3.9 full-time equivalent consultant clinical geneticists, 9.3 full-time equivalent genetic counsellors and 1 full-time equivalent genetic research nurse. This regional genetics service works closely with a local network of nurse-led breast cancer family history clinics which provide genetic counselling for patients at low or moderate familial breast cancer risk. The service hosts a genetics practice development unit promoting collaborative working between medical staff, genetic counsellors, administrators and management [26]. The study describes the usual care pathways (apart from the administration of the evaluation questionnaires) present in this service at the time.

Autonomous practice is defined in this study as 'a genetic counsellor working as part of a multi-disciplinary team, seeing patients independently and taking personal accountability for their actions'. The study was approved as a service evaluation by the hospital Research and Development office. **Participants.** The study included new patients referred to the service between the 12th December 2011 and 12th March 2012. This period was chosen pragmatically due to funding constraints. A consecutive series of 650 new patient referrals (i.e. 650 patients) were followed up until discharge (end of care episode) or until the 12th March 2013. If the referral was for a child, this counted as one patient for this study. The parents were usually seen by the clinical team at the same time as the child.

Referral management and triage process. A study tracking sheet was attached to each patient file throughout their episode of care and staff entered the date and outcome for

each contact. Data from this sheet was entered by the study administrator into a study database and this was used to prompt the administration of the patient reported outcome measure[24] and patient satisfaction questionnaire[25]. Data validation was undertaken by cross checking the study database information with patient notes and with the hospital patient information system. Outcomes included: patient group (paediatric, prenatal, adult non-cancer and adult cancer), allocation of health professional case lead, triage allocation, type of initial clinical contact offered and were formal appointments made between the hospital administration and the patient (telephone, face to face in clinic, home visit) and number of contacts per care episode.

The referral letter was reviewed by a two person on-call team consisting of a clinical geneticist and a genetic counsellor who independently completed a referral management form (supplementary material 1). The decision making process included whether to accept or decline the referral. Decline options included: decline with reasons; write back for more information or send the referral letter to another healthcare service provider. Accepted referrals were allocated by the above on-call team to either genetic counsellor or consultant geneticist led care. Any discrepancies in allocations were discussed enabling both professional viewpoints to be expressed.

The genetic counsellor then allocated an initial clinical contact at either a telephone clinic, face to face appointment in a clinic setting or home visit according to a set of existing initial clinical contact guidelines (ICCG). These guidelines were developed prior to the study by the genetic counsellor clinical team and considered the psychological, medical and social factors supplied in the referral letter. The referral was allocated one of 20 triage allocation categories

which then determined the type of initial clinical contact offered to the patient (supplementary material 2). The patient was sent a letter and a brochure explaining genetic counselling to inform their decision whether or not to opt into the service. This is standard practice in the service (supplementary material 3 and 4).

Patient questionnaires. The study aimed to determine if two self-reported questionnaires could be integrated into routine care and be used to evaluate the service. The Genetic Counselling Outcomes Scale – 24 (GCOS-24) was developed for use as a patient reported outcome measure (PROM) within clinical genetics services and has been shown to be valid, reliable and sensitive to change. The GCOS-24 score measures change in emotional, cognitive, decisional and behavioural control, as well as emotional regulation and hope[24]. The Zellerino seven item questionnaire was used to assess patient satisfaction after using clinical genetic services, this has been shown to have face validity and good internal consistency[25]. The GCOS-24 was mailed to patients before and after clinical contact. The satisfaction questionnaire was mailed after each contact. No questionnaire reminders were sent.

Results

Referral management and triage. Figure 1 shows the patient pathways followed by 650 new patients. One hundred and nine referrals (109/650, 16.8%) did not meet the referral guidelines for the service and a letter was sent back to the referrer. In some of these cases alternative service providers were suggested such as nurse-led breast cancer family history clinics. All of the remaining 541 patients were sent a letter asking them to contact the hospital administration to schedule an initial clinical contact. Eighty (14.8%) chose not to contact the service to make an appointment. This left 461 patients who were accepted by the service and

189 who decided to opt-in, to whom 825 clinical contacts were offered 7.4% (61/825) of which
190 were not attended .Types of patients included 36.3% paediatric (236/650), 4.5% prenatal
191 (29/650), 23.8% adult non-cancer (155/650) and 35.4% adult cancer (230/650).
192 Figure 1 here.
193 **Lead health professional.** Approximately half the patients, 47.8% (203/461) were triaged to
194 a genetic counsellor as the professional lead. The majority of patients (432/461, 93.7%),
195 including those triaged to clinical geneticist lead, were seen initially by a genetic counsellor.
196 For 26.8% (106/395) of the patients who attended this was their only contact with the service,
197 with the genetic counsellor able to complete the episode of care and discharge the patient.
198 Genetic counsellors contributed to 95% (784/825) of the total contacts; a clinical geneticist
199 was present in 26.9% (222/825). Only 5% (41/825) of contacts were with a clinical geneticist
200 working alone in clinic compared to 65.7% (542/825) for genetic counsellors working alone in
201 clinic.
202
203 Table 1 shows the decision outcome for triage allocation to lead professional and type of
204 initial clinical contact offered according to the patient categories included within the initial
205 clinical contact guidelines (ICCG). The categories which were more often ($p < 0.005$) triaged to
206 clinical geneticist lead included; 'where a member of the patients family had previously been
207 seen and information known' (GC: 5/25 20%, CG: 20/25 80%), 'paediatric patients with and
208 without developmental delay' (GC: 5/35 14.3%, CG: 30/35 85.7%), 'out-reach clinics' (GC: 5/20
209 25%, CG:15/20 75%) and 'where there was a new significant diagnosis in the family' (GC: 2/22
210 9.1%, CG: 20/22 90.9%). Those more often ($p < 0.001$) triaged to genetic counsellor lead
211 included 'inherited cardiac diseases' GC: 29/34 85.3%, CG: 5/34 14.7%), 'cancer predictive

testing' (GC: 30/40 75%, CG: 10/40 25%) and 'any adult referral which did not fall into another category' (GC: 87/126 69%, CG:39/126 31%).

Range and type of clinical contacts. Table 1 shows that the initial contact type offered matched well with those dictated by the initial clinical contact guidelines (figure 1). For 426 of the 461 (for whom triage allocation patient category and initial contact details were available) 39.3% were offered a telephone clinic (TC) with a genetic counsellor, 42% a hospital face to face clinic (F2F) with a genetic counsellor, 13.2% a home visit (HV) with a genetic counsellor and 4.9% a medical clinic with a clinical geneticist.

Table 1 here.

Table 2 shows the proportion of co-counselling, where either two genetic counsellors or a genetic counsellor and a clinical geneticist were present with both contributing to the counselling process for the duration of the patient consultation. The overall rate of co-counselling in this study was 29.3% (242/825), with the genetic counsellor and clinical geneticist co-counselling in 21.9% and two genetic counsellors co-counselling in 7.4% of consultations. The latter mainly represents genetic counsellor-led predictive testing for cancer and cardiac conditions. Overall, 86% (345/400) of patients who attended their appointments completed their episode of care and were discharged after one or two appointments. Only 6.7% (55/825) and 1.9% (16/825) of total contacts received third and fourth appointments.

Table 2 here.

Patient questionnaires. Developing a robust process which allowed integration of evaluation questionnaires into routine service delivery proved problematic. This stemmed from

problems with patient identification at baseline and the end of care. Only 388 out of a possible 850 satisfaction questionnaires were sent out. Ninety-seven were received (response rate 25%). This selected group of patients were satisfied by the care received by their health professional with greater than 80% stating that they 'agree somewhat' or 'strongly agree' to six out of seven questions covering; information provided, time spent, ability to answer questions, listening and engaging the patient as a partner in planning care (Figure 2).

Implementation of the questionnaires in a clinical setting was challenging. Issues identified included: a centralised clinic administration system shared with other departments and no additional resources to compensate for administration time identifying study cohort patients during the follow up period. Difficulties tracking follow up patients with the hospital patient management system meant that a reliable estimate of questionnaires sent and response rate was not possible. Only 38 pre- and post-genetic counselling GCOS-24 questionnaires were available for analysis. Those analysed showed a statistically significant difference between baseline and follow-up score (mean baseline score = 106.5, mean post-care episode score 116.1, p = 0.007 df 37), indicating that this sub-set of patients reported significant benefits from their contact with the genetics service[27].

Figure 2 here.

Discussion

The proportion of patients allocated to clinical geneticist or genetic counsellor care.

The genetic counsellors in this study led patient care for a range of genetic conditions. However, they did not take the lead for paediatric patients with developmental delay or patients with new significant diagnoses. The service did not have a 'specific' list of genetic diagnoses which were suitable for genetic counsellor led care; often this allocation was

dependent on the information provided in the individual patient referral letter. The decision making process seemed to be driven by whether there was evidence of diagnostic uncertainty or a complex medical phenotype (allocation to clinical geneticist lead) or psychological or social issues in a family with a known diagnosis (allocation to genetic counsellor lead). This supports evidence from Skirton *et al*.'s systematic review of current non-medical genetic counsellor practice[10]. This found that the majority of genetic counsellors working in other countries did not autonomously counsel cases where the diagnosis was uncertain or there was a need for a clinical examination[10].

The genetic counsellors in this study provided 73% (603/825) of the total offered contacts, as either genetic counsellor only contacts or genetic counsellor co-counselled contacts. There is similar evidence from the US where one study reports the motivation to initiate an independent genetic counsellor clinic was to relieve the pressure on the clinical geneticist's clinic[28]. In that study, 80% of the 321 patients seen did not need an additional appointment with a geneticist. In Australia, a descriptive retrospective analysis of 4817 cases saw that 42% of sessions in one region were conducted by the genetic counsellor alone[29]. In South Africa genetic counsellors independently saw 39% of the 3365 referred patients covering 57 different diagnoses[30]. The current study did not explore the health professional allocation decision making process in-depth and this is a potential area for further research.

The range of Clinical Contacts

In only 5% (41/825) of contacts did medical clinical geneticists provide counselling by themselves. The 55.9% (258/461) of the patients who required a clinical geneticist led consultation in this service would have usually been seen independently by a genetic

counsellor prior to scheduling a 2nd clinical contact as a genetic counsellor/clinical geneticists co-counselling contact. However the rates of clinical geneticist and genetic counsellor co-counselling contacts in this regional genetics service were higher than the National average at 21.9% (181/825) vs 9.9% recorded by the NHS Medical Genetics English Winter (Q3) 2013 Dashboard National Average Figures (personal communication Chair Medical Genetics Clinical Reference Group). There is currently variation in the rate of co-counselling between different genetic services in England. Possible reasons for such a high rate of clinical geneticist/genetic counsellor co-counselling in this service included the teams focus on providing a continuum of care for the patient aiming to maintain contact with the same genetic counsellor prior to, during and post clinic contacts.

Co-counselling has been one of many historical service delivery models in the UK. Co-counselling provides a multi-disciplinary approach to the holistic care of the patient and the opportunity for counsellors to focus and address different clinical and psychosocial aspects within the same consultation. Co-counselling also has benefits for the workforce as a learning opportunity for junior staff or staff expanding their scope of practice to include new areas of practice. However there are dis-advantages of co-counselling, such as the increased expense of two health professionals being present and complexities in scheduling appointments.

Genetic counsellor only contacts were higher in this study at 73.1% than the English average of 44.6%. At the time of this study the service had one of the lowest proportions of medical clinical geneticists per population covered compared to the other 19 English RGS. This required the development of effective time management and support from medical clinical geneticists for joint case review enabling genetic counsellors in this service to independently

counsel for a wide range of conditions (e.g. high risk cancers, cardiac conditions, and Huntington disease).

In this study, 29.1% (189/650) of referrals were not offered a clinical contact, representing indirect patient care which is not directly obvious from clinic attendance figures and could be overlooked when assessing workload. Of these, 94 were not accepted into the service based on the English national service specification of appropriate referrals[13]. Many of these were patients deemed to be at moderate risk of cancer which were re-directed to a network of nurse-led family history screening clinics based within oncology services. Patients re-directed to these services saw a 'specialism-specific genetic nurse' (SSGN), a registered nurse working at a specialist level of practice in a clinical specialism but with additional genetics training. A UK evaluation has shown these roles to be acceptable to patients and effective in terms of improving patient access to genetic information and tests[31 32 33].

This study is novel as it provides the first empirical evidence of the contribution of genetic counsellors within a UK regional genetic service multi-disciplinary team. Study limitations include that it describes the experience of a cohort of incoming referrals for a period of only 3 months in one centre. The practice of this one service may not be representative of other services across the UK.

Use of Initial Clinical Contact Guidelines (ICCG)

The anticipated reduction in the proportion of home visits from 26% in 2010/2011 to the 13% seen in this study (2011/2012) is thought to be a consequence of implementing revised initial clinical contact guidelines. The revised guidelines allowed for home visits to be standardised

across the team and were provided appropriately. The study demonstrated that it was possible using the guidelines to allocate patients into differing initial contact types based on the requirements of the specific case. However, this individualised process was difficult to merge with inflexible clinic booking systems and resulted in many separate clinic queues, increasing waiting times due to patients waiting for an initial telephone clinic 'slot' when other 'appointment slots' were free in other clinics. Of concern were the 21.9% (181/825) of contacts which were co-counselled by both a genetic counsellor and clinical geneticist, due to the time commitment required by both health professionals.

Moving Forwards

A decision was made to plan a service re-design to move towards a 'one clinic fits all' system. In this model genetic counsellors and clinical geneticists have separate clinics, booking is on a first come, first served basis. It is irrespective of the clinical condition or whether it is an initial or subsequent contact. The clinician still reserves the ability to perform a telephone counselling session within the allocated clinic slot if that would best meet the needs of the patient. For some patients a telephone contact to discuss concerns and facilitate a confirmation of diagnoses is thought to be most efficient. If the referral letter raises any specific psychosocial concerns, then the genetic counsellor can still ask for a co-counselling consultation to occur (including for predictive testing).

There is on-going debate as to how to most efficiently gather family history information, an initial genetic counsellor consultation usually consists of many other elements than purely information gathering. In some services the information gathering is done by specially trained administrative staff – releasing genetic counsellor time to focus on psychosocial and educative issues. One requirement of the 'one clinic fits all' model is that each genetic

356 counsellor and clinical geneticist are able to provide counselling for all paediatric, pre-natal,
357 adult and cancer genetic conditions. This is not always possible and relies on a workforce that
358 has access to and time for continuing professional development.
359
360 A framework for implementation of genetic services has recently been proposed by Rigter *et*
361 *al.* to provide structure for the transition process towards new ways of working. They defined
362 three influencing factors: different ways of doing, different ways of thinking and different
363 ways of organising [34]. The authors proposed a process of deepening, broadening and scaling
364 up of any new service. This framework could be used to help implement future service re-
365 design.
366
367 There remains the need to evaluate the effectiveness of new service designs and how the
368 limited workforce of clinical geneticists and genetic counsellors can be utilized to increase
369 access to genetic services and provide safe patient care. Currently many European countries
370 have limited or no non-medical genetic counsellors, although with increased demand for
371 access to specialist knowledge the cost effectiveness of genetic counsellors over clinical
372 geneticists may become attractive. This study has demonstrated that appropriately trained
373 and resourced genetic counsellors engaging in multi-disciplinary working practice can provide
374 a significant proportion of patient contacts. Studies undertaken in other areas of healthcare
375 could inform the optimum use of skill mix and multidisciplinary practice. The development of
376 the specialism specific genetic nurse (SSGN) role within mainstream clinical specialty areas
377 such as oncology, cardiology or endocrinology adds another route through which patients
378 could access genetic services. Cohen *et al.*[35] call for a shared terminology when describing
379 genetic service delivery models. However comparison of service delivery models between and

within countries remains difficult due to the influence of social, political and historical factors[36,37]. In the US a recent survey shows wide variation in genetic counsellor service delivery models, unfortunately in part this variation is explained by limits imposed by billing and bureaucracy, not always by good practice[38].

The methodological techniques of implementation science[39], including the conduct of implementation trials and the study of complex interventions could be beneficial in building the evidence base on which to develop new ways of working to optimise skill mix and workforce utilization in genetic service delivery models[40]. Further research needs to be undertaken to establish how best to integrate patient reported outcome and satisfaction measures within routine genetic health service delivery.

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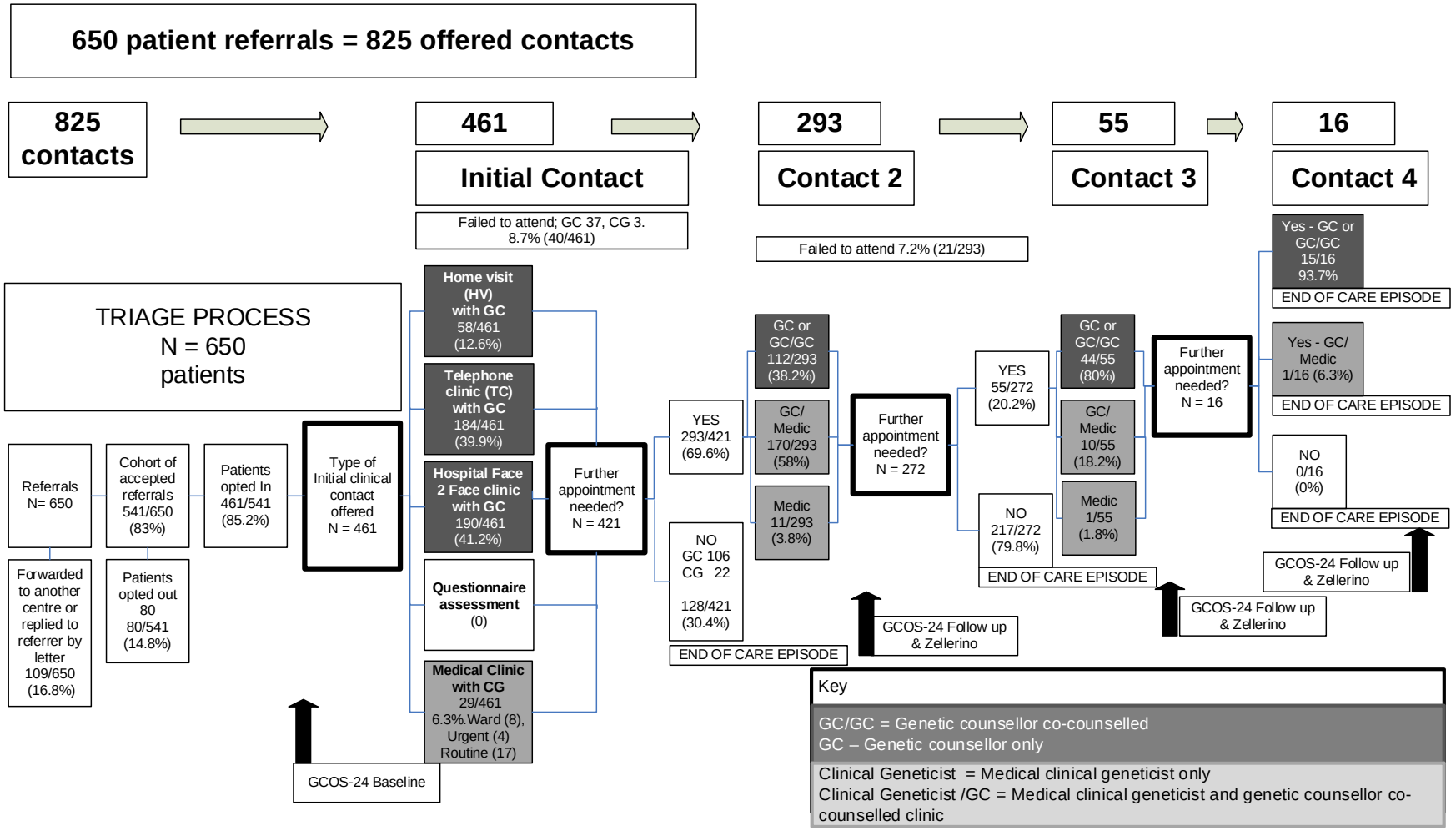
Conflict of Interest Statement: - The authors declare no conflict of interest.

Figure 1. Progress of the 650 new patients referred over the 3 month period (12.12.2011- 12.03.2012), resulting in 825 offered contacts (12 months of follow up until 12.03.2013)



423 Table 1 - Triage allocation categories for the 461 new patients who were accepted by the regional genetics service and who opted into the service.

		Lead Health Professional				Initial clinical contact *allocated*				**Total**
	Genetic counsellor Initial clinical contact advised by triage guidelines (ICCG).	Genetic Counsellor (GC)	Clinical Geneticist (CG)	Total	*P value	Telephone Clinic (TC) with GC	Face to Face (F2F) Clinic with GC	Home Visit (HV) with GC	Medical Clinic (CG) or (CG plus GC)	Total (% within patient allocation category)
Triage allocation patient category.		N (%)	N (%)	N		N (%)	N (%)	N (%)	N (%)	N (%)
All other adult referrals not mentioned below *	F2F	87 (69.0)	39 (31.0)	126	< 0.001	23 (18.1)	101 (79.6)	1 (0.8)	2 (1.6)	127 (29.8)
Inherited cardiac conditions clinic *	F2F	29 (85.3)	5 (14.7)	34	< 0.001	4 (2.4)	30 (20.7)	0 (0.0)	0 (0.0)	34 (8.0)
Predictive testing not cancer	F2F	8 (80.0)	2 (20.0)	10	0.109	1 (0.6)	9 (5.0)	0 (0.0)	0 (0.0)	10 (2.3)
Out-reach clinics with local face to face clinic *	F2F	5 (25.0)	15 (75.0)	20	0.041	5 (2.9)	12 (6.7)	2 (3.6)	1 (4.8)	20 (4.7)
Cancer predictive test *	Telephone	30 (75.0)	10 (25.0)	40	0.002	32 (18.8)	6 (3.4)	1 (1.8)	1 (4.8)	40 (9.4)
Confirmatory diagnostic testing	Telephone	15 (51.7)	14 (48.3)	29	1.000	23 (13.5)	4 (2.2)	1 (1.8)	1 (4.8)	29 (6.8)
Prenatal	Telephone	10 (33.3)	20 (66.7)	30	0.099	25 (14.7)	3 (1.7)	0 (0.0)	2 (9.5)	30 (7.0)
Paediatric patient without developmental delay *	Telephone	5 (14.3)	30 (85.7)	35	< 0.001	27 (15.9)	3 (1.7)	1 (1.8)	4 (19.0)	35 (8.2)
Family members previously seen by the service and information known *	Telephone	5 (20.0)	20 (80.0)	25	0.004	17 (10.0)	3 (1.7)	1 (1.8)	4 (19.0)	25 (5.9)
Patient lives on the Isle of Man (>50 miles)	Telephone	3 (75.0)	1 (25.0)	4	0.625	3 (1.8)	1 (0.6)	0 (0.0)	0 (0.0)	4 (0.9)
Any concerns regarding the safety of offering a home visit	Telephone	0 (0.0)	1 (100.0)	1	1.000	0 (0.0)	0 (0.0)	1 (1.8)	0 (0.0)	1 (0.2)

Terminal care issue	Home visit	2 (66.7)	1 (33.3)	3	1.000	2 (1.2)	1 (0.6)	0 (0.0)	0 (0.0)	3 (0.7)
New significant diagnosis for the family *	Home visit	2 (9.1)	20 (90.9)	22	<0.001	2 (1.2)	1 (0.6)	18 (32.1)	1 (4.8)	22 (5.2)
Paediatric patient with hearing loss	Home visit	1 (100.0)	0 (0.0)	1	1.000	0 (0.0)	1 (0.6)	0 (0.0)	0 (0.0)	1 (0.2)
Paediatric patient with life threatening illness	Home visit	1 (50.0)	1 (50.0)	2	1.000	0 (0.0)	0 (0.0)	2 (3.6)	0 (0.0)	2 (0.5)
Paediatric with developmental delay *	Home visit	0 (0.0)	32 (100.0)	32	<0.001	4 (2.4)	2 (1.2)	23 (41.1)	3 (14.3)	32 (7.5)
Evidence of parental anxiety	Home visit	0 (0.0)	1 (100.0)	1	1.000	0 (0.0)	0 (0.0)	1 (1.8)	0 (0.0)	1 (0.2)
Recent bereavement	Home visit	0 (0.0)	5 (100.0)	5	0.063	1 (0.6)	0 (0.0)	4 (7.1)	0 (0.0)	5 (1.2)
Adult with significant learning difficulties	Home visit	0 (0.0)	0 (0.0)	5 0	0.063 n/a	1 (0.6) 0 (0.0)	2 (1.2) 0 (0.0)	0 (0.0) 0 (0.0)	2 (9.5) 0 (0.0)	5 (1.2) 0 (0.0)
Reply by letter +		0 (0.0)	5 (100.0)	5	0.063	1 (0.6)	2 (1.2)	0 (0.0)	2 (9.5)	5 (1.2)
Missing category		10 (27.8)	26 (72.2)	36						
Sub-total		213 (46.2%)	248 (53.8%)			170 (39.3)	179 (42.0)	56 (13.2)	21 (4.9)	426 (100) missing 35
Total		**213**	**246**	**461**						**461**

Initial clinical contact types.

F2F **= 1 hour clinic appointment with only a genetic counsellor at health centre or hospital.**

Telephone **= 45 minutes – 1 hour booked telephone appointment with only a genetic counsellor.**

Home visit **= 1 hour appointment where the genetic counsellor would travel to the patients home (most patients lived within one hour drive from the service base).**

+ These 5 patients are included here as they were initially triaged as reply by letter but were subsequently offered an appointment.

* **Using a 5% significance level without adjustment for multiple testing for identifying triage decision where there is a significant bias in preference for either genetic counsellor or clinical geneticist lead.**

ICCG = Initial clinical contact triage guidelines

GC = UK non-medical genetic counsellor

CG = UK Medical clinical geneticist

Table 2 – Number of patient contacts offered to the 461 patients opting into the service.

	Number of patient contacts *offered* to the 461 patients.				
Health Professionals Present	**Initial Contact**	**Contact 2**	**Contact 3**	**Contact 4**	**Total**
Genetic Counsellor Only	432 (93.7%)	74 (25.3%)	25 (45.4%)	11 (68.8%)	542 (65.7%)
Genetic Counsellor/Genetic Counsellor co-counselled	0 (0.0%)	38 (13.0%)	19 (34.6%)	4 (25.0%)	61 (7.4%)
Clinical Geneticist/Genetic Counsellor co-counselled	0 (0.0%)	170 (58.0%)	10 (18.2%)	1 (6.2%)	181 (21.9%)
Clinical Geneticist only	29 (6.3%)	11 (3.7%)	1 (1.8%)	0 (0.0%)	41 (5.0%)
Total	**461 (55.9%)**	**293 (35.5%)**	**55 (6.7%)**	**16 (1.9%)**	**825 (100.0%)**

Figure Legend(s):

Table 1 - Triage allocation categories for the 461 new patients who were accepted by the regional genetics service and who opted into the service.

Table 2 - Number of patient contacts offered to the 461 patients who opted into the service.

Figure 1 – Progress of the 650 new patients referred over the 3 month period (12.12.2011-12.03.2012), resulting in 825 offered contacts – with 12 months of follow up until 12.03.2013.

Figure 2 – Patient satisfaction with the genetics service as measured by the Zellerino satisfaction questionnaire (97/388).

Supplementary material

1. Referral management form September 2011
2. Triage category sheet 2012 (ICCG) Initial Clinical Contact Guidelines.
3. Genetic Counselling Brochure sent to patients following referral to the service.
4. Hospital out-patient clinic letter inviting patients to opt-into the service.
5. New referral management form 2014

References

1. Evans D, Barwell J, Eccles D, Collins A, Izatt L, Jacobs C *et al:* The Angelina Jolie effect: how high celebrity profile can have a major impact on provision of cancer related services. *Breast Cancer Research* 2014; **16:** 442.

2. Goldsmith L, Jackson L, O'Connor A, Skirton H: Direct-to-consumer genomic testing from the perspective of the health professional: a systematic review of the literature. *Journal of Community Genetics* 2013; **4:** 169-180.

3. Schwartz MD, Valdimarsdottir HB, Peshkin BN, Mandelblatt J, Nusbaum R, Huang, A, et *al*: Randomized Noninferiority Trial of Telephone Versus In-Person Genetic Counseling for Hereditary Breast and Ovarian Cancer. *Journal of Clinical Oncology* Advance on line publication 21st January 2014. doi 10.1200/jco.2013.51.3226.

4. Iredale R, Jones L, Gray J, Deaville J: 'The edge effect': an exploratory study of some factors affecting referrals to cancer genetic services in rural Wales. *Health & place* 2005; **11:** 197-204.

5. McCann E, Baines EA, Gray JR, Proctor AM: Improving service delivery by evaluation of the referral pattern and capacity in a clinical genetics setting. *Am J Med Genet C Semin Med Genet* 2009; **151C(3):** 200-206.

6. Delikurt T, Williamson G, Anastasiadou V, Skirton H: A systematic review of factors that act as barriers to patient referral to genetic services. *European Journal of Human Genetics* 2014; Advance online publication doi: 10.1038/ejhg.2014.180.

7. Mikat- Benjamin C: Cochrane (EPOC) Title Registration. Interventions ti improve patient access, service utilization and cost of providing genetic counselling services. http://summaries.cochrane.org/title/interventions-to-improve-patient-access-service-utilization-and-cost-of-providing-genetic-counselling-services. Accessed December 2014.

8. Miller-Keane: *Definition of Healthcare Teams*, 7th edn. http://medical-dictionary.thefreedictionary.com/team: Elsevier, 2003. Accessed December 2014.

9. Kerr B, Turnpenny P, Bromillow G on behalf of the Association of Genetic Nurses and Counsellors and Clinical Genetics Society: Professional Roles in the Multidisciplinary Team in Clinical Genetics. A Framework for Practice prepared by a Working Party of the Association of Genetic Nurses and Counsellors and Clinical Genetics Society. http://www.clingensoc.org/media/43550/wp_agnc_cgs_v2.pdf Accessed December 2014: 2011.

10. Skirton H, Cordier C, Ingvoldstadt C, Taris N, Benjamin C: The role of the genetic counsellor: a systematic review of research evidence. *European Journal of Human Genetics* 2014.Advanced publication on line11th June 2014. doi:10.1038/ejhg.2014.116

11. Cordier C, Lambert D, Voelckel MA, Hosterey-Ugander U, Skirton H: A profile of the genetic counsellor and genetic nurse profession in European countries. *Journal of Community Genetics* 2012; **3:** 19-24.

12. Skirton H, Cordier C, Lambert D, Hosterey Ugander U, Voelckel M-A, O'Connor A: A study of the practice of individual genetic counsellors and genetic nurses in Europe. *Journal of Community Genetics* 2013; **4:** 69-75. https://www.eshg.org/408.0.html.Accessed December 2014.

13. NHS England: Specialised Services National Definitions Set - Medical Genetics E01/S/a. http://www.england.nhs.uk/wp-content/uploads/2013/06/e01-med-gen.pdf: 2013. Accessed December 2014.

14. NHS England Clinical Reference Group Medical Genetics: Outline Specification for the Specialist Commissioning of Genetic and Genomic Services for England. 2014. http://www.bsgm.org.uk/media/883258/bshg_july_51.pdf p 22-25. Accessed December 2014.

15. Barnes C, Kerzin-Storrar L, Skirton H, Tocher J: The Department of Health-supported genetic counsellor training post scheme in England: a unique initiative? *J Community Genet* 2012; **3:** 297–302.

16. Association of Genetic Nurses and Counsellors: Results from joint AGNC/lead genetic counsellor meeting 20 October 2011. Available from the Chair of the Association of Genetic Nurses and Counsellors.

17. Skirton H, Kerzin-Storrar L, Barnes C, Hall G, Longmuir M, Patch C *et al*: Building the genetic counsellor profession in the United kingdom: two decades of growth and development. *Journal of Genetic Counseling* 2013; **22:** 902-906.

18. EURORDIS Rare Diseases Europe, Kole A, Faurisson F: The voice of 12,000 patients. Experiences and Expectations of Rare Disease Patients on Diagnosis and Care in Europe. A report based on the EurordisCare2 and EurordisCare3 Surveys. 2009. http://www.eurordis.org/publication/voice-12000-patients. Accessed December 2014.

19. Rare Disease UK: Experiences of Rare Diseases: An insight from Patients and Families. http://www.raredisease.org.uk/documents/RDUK-Family-Report.pdf, 2010.Accessed December 2014.

20. Burton H, Cole T, Farndon P: *Genomics in medicine - delivering genomics through clinical practice.* Public Health Genetics Foundation, 2012.

21. Laurant M, Reeves D, Hermens R, Braspenning J, Grol R, Sibbald B: Substitution of doctors by nurses in primary care. *Cochrane Database of Systematic Reviews* 2005.

22. Richardson G: Identifying, evaluating and implementing cost-effective skill mix. *Journal of Nursing Management* 1999; **7:** 265-270.

23. Dierick-van Daele ATM, Spreeuwenberg C, Metsemakers JFM, Vrijhoef BJM: Critical appraisal of the literature on economic evaluations of substitution of skills between professionals: a systematic literature review. *Journal of Evaluation in Clinical Practice* 2008; **14:** 481-492.

24. McAllister M, Wood A, Dunn G, Shiloh S, Todd C: The Genetic Counselling Outcome Scale: a new patient reported outcome measure for clinical genetics services. *Clinical Genetics* 2011; **79:** 413-424.

25. Zellerino B, Milligan SA, Brooks R, Freedenberg DL, Collingridge DS, Williams MS: Development, testing, and validation of a patient satisfaction questionnaire for use in the clinical genetics setting. *American Journal Of Medical Genetics Part C, Seminars In Medical Genetics* 2009; **151C:** 191-199.

26. Norris A: Practice development unit - every person makes a difference. *BMJ supportive & palliative care* 2011; **1:** 258-259.

27. McAllister M, Payne K, Nicholls S, MacLeod R, Donnai D, Davies L: Patient empowerment in clinical genetics services. *J Health Psychol* 2008; **13:** 887-897.

28. Hannig V, Cohen M, Pfotenhauer J, Williams M, Morgan T, Phillips Jr: Expansion of Genetic Services Utilizing a General Genetic Counseling Clinic. *Journal Of Genetic Counseling* 2013; **23** (1): 64-71.

29. Kromberg JGR, Parkes J, Taylor S: Genetic Counselling as a developing Healthcare Profession: A case study in the Queensland Context. *Australian Journal of Primary Health* 2006; **12:** 33-39.

30. Kromberg JGR, Wessels TM, A K: Roles of genetic counsellors in South Africa. *Journal of Genetic Counselling* 2013; **6:** 753-761.

31. Kirk M, Simpson A, Llewellyn M, Tonkin E, Cohen D, Longley M: Evaluating the role of Cardiac Genetics Nurses in inherited cardiac conditions services using a Maturity Matrix. *European Journal of Cardiovascular Nursing* 2014; **13**:5:418-428. Advanced on line publication 2013; doi: 10.1177/1474515113502748.

32. Martin G, Weaver S, Currie G, McDonald R, Finn R: Innovation sustainability in challenging health-care contexts: embedding clinically led change in routine practice. *Health Services Management Research* 2012; **25:** 190-199.

33. Bennett CL, Burke SE, Burton H, Farndon PA: A toolkit for incorporating genetics into mainstream medical services: Learning from service development pilots in England. *BMC Health Services Research* 2010; **10**.125.

34. Rigter T, Henneman L, Broerse J, Shepherd M, Blanco I, Kristoffersson, U *et al*: Developing a framework for implementation of genetic services: learning from examples of testing for monogenic forms of common diseases. *J Community Genet* 2014; **5:** 337-347.

35. Cohen SA, Marvin ML, Riley BD, Vig HS, Rousseau JA, Gustafson SL: Identification of genetic counseling service delivery models in practice: a report from the NSGC Service Delivery Model Task Force. *Journal of Genetic Counseling* 2013; **22:** 411-421.

36. Cohen S, Gustafson S, Marvin ML, Vig HS, Rousseau, J A, Gustafson, S L *et al*: Report from the National Society of Genetic Counselors service delivery model task force: a proposal to define models, components, and modes of referral. *Journal of Genetic Counseling* 2012; **Oct;21(5):** 645-651.

37. Battista RN, Blancquaert I, Laberge AM, van Schendel N, Leduc N: Genetics in health care: an overview of current and emerging models. *Public Health Genomics* 2012; **15:** 34-45.

38. Trepanier A, D A: Models of Service delivery for cancer Genetic Risk Assessment and Counselling. *Journal of Genetic Counseling* 2014; **23:** 239-253.

39. Peters DH, Taghreed A, Alonge O: Implementation research: what it is and how to do it.: *BMJ*, 2013, Vol 347, pp 1-7.

40. Craig P, Dieppe P, Nacintyre S, Mitchie S, Nazareth I, Petticrew M: Developing and evaluating complex interventions: the new Medical Research Council guidance. *BMJ* 2008; **337:** 979-983.