

1 **Title**

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

4 **Running title** - Referral management and workforce allocation.

5 **Authors**

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26 **Abstract**

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

48

49 **Introduction**

50 Over the last few years advances in technology have resulted in a rapid increase in the number
51 of gene tests available. In parallel there has been an increase in the number of referrals to
52 genetic services due to heightened public and health professional awareness of the potential
53 of genomic information^{1,2}. Recent work indicates that workforce interventions within service
54 re-organisation; including telephone counselling by genetic counsellors³, the provision of
55 more genetic specialists in rural areas⁴ and changes to the referral management systems⁵
56 can have a significant effect on the ability of patients to access genetic service^{6,7}. A systematic
57 literature review, entitled “Interventions to improve patient access, service utilization and
58 cost of providing genetic counselling services” is currently planned⁷.

59

60 The present study describes the usual care pathways of a UK regional genetics service in 2011-
61 2012. One study aim was to monitor the use of recently introduced new initial clinical contact
62 guidelines (ICCG) with the aim of reducing the proportion of home visits and increasing the
63 proportion of telephone contacts. The service provided 26% of its initial contacts by a home
64 visit in 2010-2011.

65

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

72

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

83

84 Currently the utilization of non-medical genetic counsellors varies across Europe and many
85 countries only have a small number of practitioners¹¹. In order to safeguard the public and
86 standardise practice, the European Board of Medical Genetics has defined a code of practice
87 and registration system for genetic counsellors across Europe¹². English regional genetic
88 services receive centralised funding¹³. The services are provided free to patients at the point
89 of access. The NHS England Medical Genetics Clinical Reference Group has responsibility for
90 defining English service specifications and outcomes for the 23 regional genetics services¹⁴.
91 UK genetic counsellor professional registration is regulated by the UK and Eire Genetic
92 Counsellor Registration Board. It is based on the principle that in addition to prior academic
93 and vocational qualifications, registered genetic counsellors are required to have a 2-year
94 training period and a Masters level portfolio assessment to become appropriately
95 competent¹⁵. In 2011 there were 272 genetic counsellor positions in the UK, but not all
96 practitioners were registered with the GCRB¹⁶. The current number of practitioners is

97 unknown but 186 are UK Genetic Counsellor Board Registered. The majority working as part
98 of a multi-disciplinary team attached to regional genetics services¹⁷.

99

100 The situation across Europe is complicated by differing economic policy, healthcare
101 professional roles, cultural preferences and political systems. Patient surveys show that there
102 is great variability in access to genetic healthcare for diagnosis and on-going treatment ^{18;19}.

103 To meet this demand clinicians are moving away from established models of service delivery
104 and are developing innovative ways of working such as the integration of genetic and genomic
105 testing within mainstream clinical specialty areas²⁰. This refinement of skill mix and workforce
106 optimisation has been seen within other areas of healthcare typified by rising consumer
107 demand and limited resources²¹⁻²³. Historically the UK non-medical genetic service workforce
108 provided psychosocial support and counselling which originated in many services from a
109 community family nursing perspective. This aimed to provide on-going holistic care to families
110 affected by genetic conditions. This focus on continuity of care in a community setting has
111 meant that some services still retain the option of offering a home visit in certain
112 circumstances if indicated by local protocols. This study was designed to describe practice
113 within a regional genetics service and aimed to:

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

119

- 120 • Explore whether a genetic patient reported outcome measures questionnaire²⁴ and a
121 patient satisfaction questionnaire²⁵ can be incorporated into service provision.

122 **Methods**

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

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

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

143 each contact. Data from this sheet was entered by the study administrator into a study
144 database and this was used to prompt the administration of the patient reported outcome
145 measure²⁴ and patient satisfaction questionnaire²⁵. Data validation was undertaken by cross
146 checking the study database information with patient notes and with the hospital patient
147 information system. Outcomes included: patient group (paediatric, prenatal, adult non-
148 cancer and adult cancer), allocation of health professional case lead, triage allocation, type
149 of initial clinical contact offered and were formal appointments made between the hospital
150 administration and the patient (telephone, face to face in clinic, home visit) and number of
151 contacts per care episode.

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

160

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

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

170

171 **Patient questionnaires.** The study aimed to determine if two self-reported questionnaires
172 could be integrated into routine care and be used to evaluate the service. The Genetic
173 Counselling Outcomes Scale – 24 (GCOS-24) was developed for use as a patient reported
174 outcome measure (PROM) within clinical genetics services and has been shown to be valid,
175 reliable and sensitive to change. The GCOS-24 score measures change in emotional, cognitive,
176 decisional and behavioural control, as well as emotional regulation and hope²⁴. The Zellerino
177 seven item questionnaire was used to assess patient satisfaction after using clinical genetic
178 services, this has been shown to have face validity and good internal consistency²⁵. The GCOS-
179 24 was mailed to patients before and after clinical contact. The satisfaction questionnaire was
180 mailed after each contact. No questionnaire reminders were sent.

181 **Results**

182 **Referral management and triage.** Figure 1 shows the patient pathways followed by 650 new
183 patients. One hundred and nine referrals (109/650, 16.8%) did not meet the referral
184 guidelines for the service and a letter was sent back to the referrer. In some of these cases
185 alternative service providers were suggested such as nurse-led breast cancer family history
186 clinics. All of the remaining 541 patients were sent a letter asking them to contact the hospital
187 administration to schedule an initial clinical contact. Eighty (14.8%) chose not to contact the
188 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

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

214

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

221 Table 1 here.

222

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

233 Table 2 here.

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

236 problems with patient identification at baseline and the end of care. Only 388 out of a possible
237 850 satisfaction questionnaires were sent out. Ninety-seven were received (response rate
238 25%). This selected group of patients were satisfied by the care received by their health
239 professional with greater than 80% stating that they 'agree somewhat' or 'strongly agree' to
240 six out of seven questions covering; information provided, time spent, ability to answer
241 questions, listening and engaging the patient as a partner in planning care (Figure 2).

242

243 Implementation of the questionnaires in a clinical setting was challenging. Issues identified
244 included: a centralised clinic administration system shared with other departments and no
245 additional resources to compensate for administration time identifying study cohort patients
246 during the follow up period. Difficulties tracking follow up patients with the hospital patient
247 management system meant that a reliable estimate of questionnaires sent and response rate
248 was not possible. Only 38 pre- and post-genetic counselling GCOS-24 questionnaires were
249 available for analysis. Those analysed showed a statistically significant difference between
250 baseline and follow-up score (mean baseline score = 106.5, mean post-care episode score
251 116.1, $p = 0.007$ df 37), indicating that this sub-set of patients reported significant benefits
252 from their contact with the genetics service²⁷.

253 Figure 2 here.

254 **Discussion**

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

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

260 dependent on the information provided in the individual patient referral letter. The decision
261 making process seemed to be driven by whether there was evidence of diagnostic uncertainty
262 or a complex medical phenotype (allocation to clinical geneticist lead) or psychological or
263 social issues in a family with a known diagnosis (allocation to genetic counsellor lead). This
264 supports evidence from Skirton *et al.*'s systematic review of current non-medical genetic
265 counsellor practice¹⁰. This found that the majority of genetic counsellors working in other
266 countries did not autonomously counsel cases where the diagnosis was uncertain or there
267 was a need for a clinical examination¹⁰.

268

269 The genetic counsellors in this study provided 73% (603/825) of the total offered contacts, as
270 either genetic counsellor only contacts or genetic counsellor co-counselled contacts. There is
271 similar evidence from the US where one study reports the motivation to initiate an
272 independent genetic counsellor clinic was to relieve the pressure on the clinical geneticist's
273 clinic²⁸. In that study, 80% of the 321 patients seen did not need an additional appointment
274 with a geneticist. In Australia, a descriptive retrospective analysis of 4817 cases saw that 42%
275 of sessions in one region were conducted by the genetic counsellor alone²⁹. In South Africa
276 genetic counsellors independently saw 39% of the 3365 referred patients covering 57
277 different diagnoses³⁰. The current study did not explore the health professional allocation
278 decision making process in-depth and this is a potential area for further research.

279

280 **The range of Clinical Contacts**

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

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

294

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

302

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

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

310

311 In this study, 29.1% (189/650) of referrals were not offered a clinical contact, representing
312 indirect patient care which is not directly obvious from clinic attendance figures and could be
313 overlooked when assessing workload. Of these, 94 were not accepted into the service based
314 on the English national service specification of appropriate referrals¹³. Many of these were
315 patients deemed to be at moderate risk of cancer which were re-directed to a network of
316 nurse-led family history screening clinics based within oncology services. Patients re-directed
317 to these services saw a 'specialism-specific genetic nurse' (SSGN), a registered nurse working
318 at a specialist level of practice in a clinical specialism but with additional genetics training. A
319 UK evaluation has shown these roles to be acceptable to patients and effective in terms of
320 improving patient access to genetic information and tests^{31 32 33}.

321

322 This study is novel as it provides the first empirical evidence of the contribution of genetic
323 counsellors within a UK regional genetic service multi-disciplinary team. Study limitations
324 include that it describes the experience of a cohort of incoming referrals for a period of only
325 3 months in one centre. The practice of this one service may not be representative of other
326 services across the UK.

327

328 **Use of Initial Clinical Contact Guidelines (ICCG)**

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

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

340 **Moving Forwards**

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

350

351 There is on-going debate as to how to most efficiently gather family history information, an
352 initial genetic counsellor consultation usually consists of many other elements than purely
353 information gathering. In some services the information gathering is done by specially trained
354 administrative staff – releasing genetic counsellor time to focus on psychosocial and
355 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 organising³⁴. 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

380 within countries remains difficult due to the influence of social, political and historical
381 factors^{36,37}. In the US a recent survey shows wide variation in genetic counsellor service
382 delivery models, unfortunately in part this variation is explained by limits imposed by billing
383 and bureaucracy, not always by good practice³⁸.

384

385 The methodological techniques of implementation science³⁹, including the conduct of
386 implementation trials and the study of complex interventions could be beneficial in building
387 the evidence base on which to develop new ways of working to optimise skill mix and
388 workforce utilization in genetic service delivery models⁴⁰. Further research needs to be
389 undertaken to establish how best to integrate patient reported outcome and satisfaction
390 measures within routine genetic health service delivery.

391

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397

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

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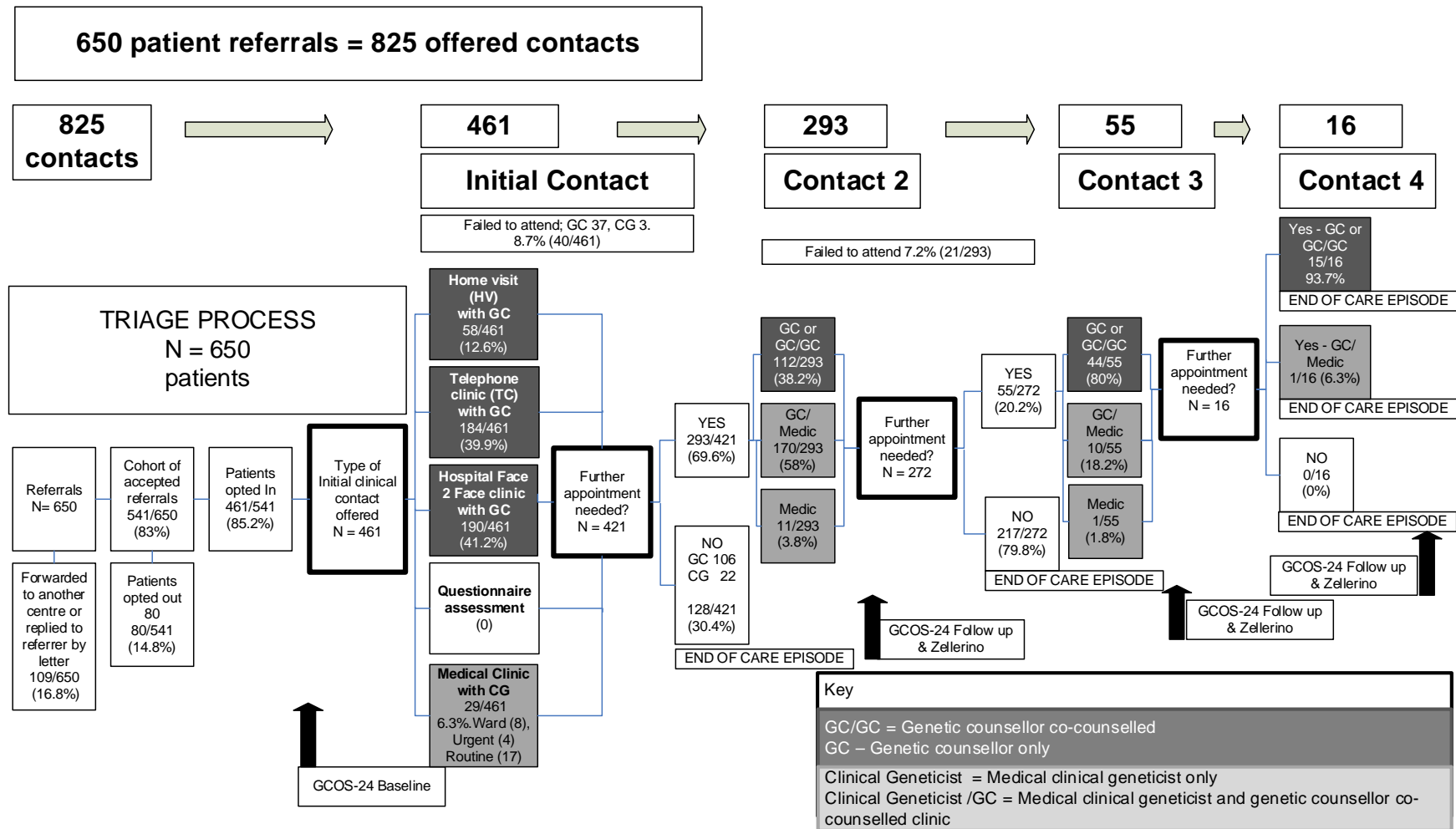
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Figure 1. Progress of the 650 new patients referred over the 3 month period (12.12.2011- 12.03.2012), resulting in 825 offered contacts (12 months of follow up until 12.03.2013)



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.

	Genetic counsellor Initial clinical contact advised by triage guidelines (ICCG).	Lead Health Professional				*P value	Initial clinical contact <i>allocated</i>				Total (%) within patient allocation category)						
		Genetic Counsellor (GC)	Clinical Geneticist (CG)	Total			Telephone Clinic (TC) with GC	Face to Face (F2F) Clinic with GC	Home Visit (HV) with GC	Medical Clinic (CG) or (CG plus GC)							
Triage allocation patient category.		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)	0	n/a	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	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

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

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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%)	

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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

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