Children with Rare Chromosome Disorders: UK families' experiences of diagnosis and counseling 2003 - 2013

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CONFLICT OF INTEREST
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ABSTRACT

Introduction: The United Kingdom (UK) Strategy for Rare Diseases places a strong emphasis on the need to empower people affected in order to improve diagnosis, intervention, and coordination of care. An important group to consider are families with children with a rare chromosome disorder (RCD). This study reports on families' experiences of diagnosis and counselling, highlights changes over ten years, and identifies recommendations for improvement.

Methods: Two national surveys were undertaken by Unique (Rare Chromosome Disorder Support Group) to compare families' experiences over the decade prior to the launch of the strategy. Questionnaires explored seven stages of the care pathway from pre-testing, to diagnosis, genetics consultation, follow-up and then sign-posting to peer support, plus measurement of perceived service quality.

Results: Response rates were: 36.4% in 2003 (583 families) and 53.6% in 2013 (575 families). Mean age of respondents was 42.3 years and 43.0 years respectively. Analysis of responses identified 28 user-identified areas for service improvement for families affected by RCDs. Only 12/28 are currently incorporated in service specifications.

Conclusions: Identification of user-led, evidence-based recommendations can empower those affected by RCDs and enable professionals to co-design improved services through involvement of family support groups. A series of further surveys is planned.

Key Words: Families' Experiences, Clinical Genetics Services, Rare Chromosome Disorders, National Surveys, Patient Reported Outcomes (PROs), Evidence-Based Clinical Guidelines.

1. INTRODUCTION

The United Kingdom (UK) Strategy for Rare Diseases places a strong emphasis on empowering those affected by rare diseases in order to improve diagnosis, intervention, and coordination of care in genetics services [1]. Although the strategy was published in late 2013, implementation plans are still being developed for England in 2017 with a view to being fully actioned in 2020. It is acknowledged that this will require "strengthening the mechanisms and opportunities for meaningful and sustained patient involvement in rare disease service provision".

An important and challenging group to consider when involving those affected by rare diseases will be children with a rare chromosome disorder (RCD). In the UK, it is estimated that at least 300-500 children are born every year with one of a range of RCDs, widely spread geographically [2]. In comparison to more common and well-studied chromosome disorders like Down syndrome, there is far less information available on the natural history or prognosis for these rare diseases (<5 per 10,000 births) [3]. Their extreme rarity means RCD cases can be particularly challenging for genetics services because, in addition to communicating a laboratory diagnosis, professionals also need to support families who frequently experience severe distress combined with high levels of uncertainty [4]. In such a situation, service providers must ensure that parents understand the diagnosis, help families identify effective coping strategies, and address the lack of available evidence [5 6]. To date, little is known about the experiences of these families, or the degree to which Clinical Genetics Services currently meet their needs. This is an important gap, since the UK strategy emphasises that successful implementation will require "recognising patient groups as key partners" to develop care pathways that incorporate "best practice from the user perspective" [1].

Patient-reported outcomes for Clinical Genetics Services are still in their infancy [7]. Over the last decade some developments have occurred, largely driven by the extension of clinical genetic services from diagnosing conditions that are exclusively genetic in nature to investigating genetic components for more common diseases [8], with increased knowledge about the contribution of genetic factors to a range of common diseases [9]. Comprehensive data are not yet available for RCDs although it is anticipated that, in time, the new National Congenital Anomaly and Rare Disease Registration Service established by Public Health England will fill this gap in knowledge [10], with projects such as the Sanger DECIPHER database [11] and the Unique registry/database and information guide service [12] also contributing to an improved knowledge base. Currently, there is no European Reference Network (ERN) specific to RCDs despite pan-European efforts to create one. However, the UK-led ERN-Ithaca for intellectual disability and congenital malformations will include RCDs with family support group representation and promises to be a channel through which the experiences of RCD families can be improved.

For chromosome disorders, technological developments such as the introduction of microarray-based comparative genomic hybridisation (*microarray analysis*) techniques have meant that chromosome abnormalities which were formerly too small to be detected by conventional karyotyping can now be identified. Although this has significantly improved sensitivity for detection of clinically relevant genomic imbalances, it has also increased the need for comprehensive genetic counselling to ensure accurate clinical interpretation [13]. In the case of RCDs, clinical interpretation will still face a high level of uncertainty about each affected child's health, potential cognitive development, and life span even after there is a definitive diagnosis [14-16].

In this paper, we present the findings of two large scale surveys which investigated the experiences of UK families who have a child with a RCD over the period 2003 – 2013. We examined the entire care pathway including provision of pre-test information, diagnosis of RCD, genetic counselling, provision of follow-up information and ongoing support. Analysis of responses at different time-points is used to reveal trends and changes over time. The findings should hopefully enable best practice from the user perspective to be more effectively integrated into the implementation phase of the UK Strategy for Rare Diseases.

1.1 Objectives

The study had three main objectives:

1. to examine RCD families' experiences along the entire care pathway;
2. to compare differences over ten years and identify positive or negative changes over time; and
3. to recommend improvements to service provision for this important patient group.

2. METHODS

2.1 Survey Overview

Two surveys were undertaken using a detailed questionnaire designed by Unique, a UK-based Rare Chromosome Disorder Support Group. The group has over 15,300 member families, representing over 17,000 individuals affected by RCDs, in over 100 countries worldwide, with around 1500 new families registering annually [17]. The process for designing the questionnaire is described in Supplementary file 1. The first survey was undertaken in March 2003 and the second in May 2013. Both surveys were limited to members with at least one surviving child with RCD and a valid UK address, and the 2013 survey to UK families who had joined the group since March 2003. Both were identical, except for the addition of some questions in 2013 relevant to the introduction of microarray analysis [18]. The layout of questionnaires was designed to minimise the possibility of systematic missing responses. Questionnaires in 2003 were pre-printed and posted out to families while questionnaires in 2013 could be completed online or printed off and returned by post; responses were anonymous. The initial invitation in 2013 was followed by two email reminders.

2.2 Questionnaire Content

Questionnaires collected background information on the family. Respondents were then asked about their experiences during different stages of the patient journey (see Table 1). A separate question asked families to rate the quality of the overall service from a user perspective in terms of the overall service received on a ten-point Likert scale ranging from 1 (worst) to 10 (best), and how helpful overall the genetics counselling service has been since their first appointment (4 categories ranging from 'not very helpful' to 'very helpful'). Finally, respondents were invited to give free text descriptions of their experiences of diagnosis and genetic counselling. A copy of the 2003 postal questionnaire is provided in Supplementary file 2.

2.3 Data Analysis

Numerical data were summarised using mean and SD or median and range, depending on data distribution. Analysis was based on completed question responses. There was no imputation of missing data, although we investigated to assess as far as is possible that missing data were missing completely at random. Certain descriptive variables with multiple response categories were dichotomized before analysis e.g. whether person communicating diagnosis was 'genetics professional' or 'non-genetics professional', whether the method of communicating was 'in person face-to-face' or 'indirect by phone, letter etc.'. For comparison of baseline and 2013 responses, chi-square tests were performed for categorical variables and *t*-tests or Mann–Whitney *U* tests for continuous variables. 95% confidence intervals were estimated together with the significance level of observed differences. In addition, some 2013 survey responses were analysed separately for cases diagnosed before and after the introduction of microarray tests in 2008. Stata (version 13) was used for all analyses. Statistical significance was set at p=0.001 level.

2.4 Recommended Improvements

A list of recommended improvements was compiled by knowledgeable family members with direct personal experience of RCD, as well as a clinical geneticist and genetics laboratory scientists. Recommendations were based on analysis of data extracted from the questionnaire responses (with detailed examination of levels, significant changes or lack of a significant difference over time).

3. RESULTS

3.1 Respondents

A total of 583/1600 families responded to the 2003 survey (36.4% response rate). In 2013, of 584 responses received; 9 families not resident in the UK were excluded, leaving a total of 575/1072 questionnaires for analysis (53.6 % response rates).

Respondent characteristics were similar in the two groups. Mean age was 42.3 years in 2003 and 43.0 years in 2013. Questionnaires were mainly completed by mothers, although this proportion fell over time from 92.3% to 85.9% in 2013. The majority described themselves as White British/ White European, although ethnic minority respondents doubled over the period from 4.8% to 8.5%. Most families had only one child with a RCD, with this figure rising over the ten years from 86.1% to 92.3%. A small minority of families had lost a

child with a RCD at or after birth; this figure had reduced over time from 6.5% in 2003 to 2.2% in the 2013 sample.

3.2 Rating of service received

When asked how helpful the genetics counselling service had been since their first appointment, Figure 1a shows views were fairly evenly spread across the four categories ranging from 'not very helpful' to 'very helpful', although the most common response was 'had no more contact'. The percentage rating a service as 'not very helpful' did not alter over time; it was 18.7% (95% Confidence Interval (CI): 15.1 - 22.7) in 2003 and 15.0% (95% CI: 11.6 - 19.0) in 2013 (p=0.161).

When asked to rate overall service quality on a ten-point scale, average scores rose from 6.37 [SD 2.63] in 2003 to 7.00 [SD 2.52] in 2013. Figure 1b shows mean scores for different professional groups (i.e. genetics doctors, genetics counsellors, and genetics nurses). In 2013, scores were 7.1 (95% CI 6.9, 7.3), 6.5 (6.0, 7.1), and 6.6 (5.7, 7.5) respectively. Therefore, using an unpaired t-test, the genetics doctors scored statistically significantly higher than the genetics counsellors. Comparison of the genetics doctors with the nurses, and of the counsellors with nurses, were not statistically significant.

3.3 Families' experiences over ten years

Families' experiences of services over time are presented in Table 2.

1: Pre-testing process: In 2003 only 70.7% (95% CI: 66.4 - 74.7) of families reported that they had been informed that their child's chromosomes were going to be tested. In 2013, this figure was slightly higher at 73.2% (95% CI: 68.6 - 77.5) but showed no significant improvement over the ten years (p=0.404).

2: Test result communication: In 2003, test results were far more likely to be communicated by a paediatrician (64.5% (95% CI: 60.3 - 68.5)) than a genetic specialist (23.8% (95% CI: 20.2 - 27.5)). By 2013, results were almost equally likely to be communicated by a genetic specialist (49.0% (95% CI: 44.3 - 53.7)) or a paediatrician (45.0% (95% CI: 40.3 - 49.7)). Results were rarely reported by other professionals e.g. GPs, genetic nurses, obstetricians, health visitors.

Table 2 indicates that, over the ten year period, it has become significantly less likely (p<0.001) that parents will be informed in person about their child's chromosome disorder, although even in 2013 the majority still stated that they were told in person (62.7% (95% CI: 58.2, 67.1)) versus 76.1% (95% CI: 72.4 - 79.6) in 2003. During the same period, communication by telephone doubled from 12.2% (95% CI: 9.6 - 15.2) to 22.1% (95% CI: 18.4 - 26.1) in 2013, and by 50% for letters from 10.1% to 14.4%. Similar trends, away from telling parents in person, are observable for both professional groups (i.e. paediatricians and genetic specialists).

Possibly linked to this trend, responses indicate a shift towards families receiving their test result at home. This has risen significantly (p<0.001) from 17.1% (95% CI: 14.0, 20.6) in 2003 to 27.1% (95% CI: 22.9, 31.7) in 2013. At the same time, there has been an increase (p=0.006) in test results being communicated in a genetics centre from 8.7% (95% CI: 6.5 - 11.5) to 14.4% (95% CI: 11.1, 18.2); and a significant drop (p<0.001) in parents receiving information on the ward after birth or on the children's ward (28.5% (95% CI: 24.6 - 32.5) vs. 14.7% (95% CI: 11.4 - 18.4). Throughout, one in four families continued to receive their test results in a doctor's surgery (26.7% vs. 23.6% in 2013) and one in ten in a child development centre (11.3% vs. 11.5%). The proportion who are told in private has not changed significantly (p=0.697) over this period; 80.2% (95% CI: 76.5 - 83.5) in 2003 and 81.1% (95% CI: 77.2 - 84.8) in 2013. Services did not always ensure that support was available from a spouse/partner, relative or friend when imparting this life-changing information. In 2003, one quarter of respondents (23.1% (95% CI: 19.5 - 26.9)) were on their own when they received the diagnosis; rising slightly (p=0.082) to 28.0% (95% CI: 23.8 - 32.4)) in 2013. In addition, 47.3% (95% CI: 42.9 - 51.7)) said that their affected child had been present in 2003, and 46.1% (95% CI: 41.3 - 50.9)) in 2013, indicating no significant change (p=0.710).

3: Referral to a genetic specialist: Table 2 shows that the proportion of families receiving genetic counselling has decreased slightly (p=0.031) from 58.4% (95% CI: 54.2 - 62.5)) in 2003 to 52.0% (95% CI: 47.7 - 56.2)) in 2013. For families informed about their child's test result by a non-geneticist (i.e. paediatrician, GP etc.) likelihood of referral to a genetic specialist has not increased (p=0.322), with two out of ten not offered a referral; 22.3% (95% CI: 18.4 - 26.7)) in 2003 and 19.3% (95% CI: 14.9 - 24.2)) in 2013. In families where a second child was diagnosed with an RCD, this figure remains similar (22.7% vs. 21.7% in 2013).

Once referred, the waiting time for an appointment was over 3 months with a slight non-significant (p=0.105) increase over time; 95.7 days in 2003 (95% CI: 83.6 - 107.7)) and 115.0 days (95% CI: 94.4 - 135.7)) in 2013. For a medical geneticist the time has increased from 103.5 days to 172.1 days and for a genetic counsellor from 118 days to 199.1 days. However, for genetic nurses waiting times have fallen from 118 days to 75.6 days. In cases where a definitive diagnosis could not be made at the time of the appointment (15%), further tests are increasingly likely to be ordered; 43.1% of such cases in 2003 and 60.0% in 2013. These further tests produce a change in the provisional diagnosis in one in ten cases (11.6% in 2003 and 9.9% in 2013).

4: Conduct of genetic consultation: Table 2 shows that most respondents considered they had been informed of their child's condition in a sensitive manner; with a slight rise (p=0.014) from 66.9% (95% CI: 62.6 - 70.9)) in 2003 to 74.1% (95% CI: 69.8 - 78.2)) in 2013. There appear to be consistent differences between the perceived sensitivity of different professional groups; for genetic professionals, 81.6% in 2003 and 86.2% in 2013 were viewed as providing the information sensitively, 63.3% and 67.7% of paediatricians respectively, and 43.8% vs. 44.0% of other clinicians.

The conduct of consultations was explored in some detail. Although genetic specialists always introduced themselves (>98% consultations), families were not always told how long the consultation would take (45.6% vs. 38.1% in 2013) or asked what information they already had (24.7% vs. 24.3% in 2013 not asked), and half were not asked how detailed they would like information provided to be (56.5% vs. 54.4% in 2013). Almost half of respondents thought the genetic specialist did not seem to know about them and their family (49.1% vs. 42.3% in 2013), one in five said that a family genetic history was not taken (20.7% vs. 21.0% in 2013), and one third said that there had been no physical examination of their child (37.4% vs. 31.9% in 2013). The risk of having another baby with an RCD was not always explained, with evidence of a decline over time and variation between professionals; medical geneticists (11.8% vs. 20.0% in 2013 did not explain), genetic counsellors (19.0% vs. 32.1%), and genetic nurses (27.8% vs. 30.0%).

Provision of a written summary following the genetic consultation is considered to be good practice [19]. In 2003, written summaries were provided by 69.0% of medical geneticists, 50.7% of genetics counsellor and 43.8% of genetics nurses. By 2013, although figures had risen to 81.0%, 65.3% and 65.4% respectively, they were still not provided for all as routine practice. On average, families had to wait one month to receive a summary, but some waited as long as 6 months; in 2013 longer delays were reported. The written information provided was considered easy to understand by almost all recipients (93.0% in both 2003 and 2013).

5: Genetic and clinical information provision: Although most respondents could understand the information provided (92.7% in 2003 vs. 89.7% in 2013), the majority considered they had not been given enough information about their child's condition. Table 2 shows this did not change significantly (p=0.093) over time; 69.4% (95% CI: 65.2 - 73.4) in 2003 and 64.3% (95% CI: 59.6 - 68.8) in 2013. Responses were not affected by the introduction of microarray analysis, with 66.4% pre-2008 and 61.4% post-microarrays reporting a need for more information. In terms of the content of the information provided, although the majority of families were told which chromosome numbers were involved this has not increased significantly (p=0.067); 78.5% (95% CI: 74.6 - 82.0) were told in 2003 and 83.3% (95% CI: 79.4 - 86.7) in 2013. However, explanation of the type of chromosome disorder in a clear and understandable way has improved significantly (p<0.001) rising from 57.2% (95% CI: 52.8 - 61.6) in 2003 to 75.1% (95% CI: 70.7 - 79.1) in 2013. Similarly, although a significant proportion of families are not given the karyotype, this has decreased over time (44.4% in 2003 falling to 34.0% in 2013).

Virtually all respondents (95%) said they would have liked a copy of the genetics laboratory report. Although this was not provided in the majority of cases, there is evidence that families are increasingly likely to be given a copy. In 2003, 71.1% were not given a copy, compared to only 48.6% in 2013. However, when laboratory reports were provided, only half included a suitable explanation of the medical or technical terms used, with no evidence of improvement over time (52.2% in 2003 vs. 50.2% in 2013).

In terms of the clinical prognosis, nearly one in three respondents said that they were not told the possible effects on their child of the chromosomal abnormality (30.1% vs. 28.7% in 2013). In cases where this is provided, accuracy appears to have improved over time e.g. for genetic nurses from 68.3% to 75.7% considered accurate in 2013.

6: Genetic service follow-up: Although most families were offered a further meeting to discuss their child's chromosome disorder, one third reported that they were not. There was a slight but non-significant (p=0.158)

improvement over time. In 2003, 35.2% (95% CI: 31.1 - 39.5) were not offered a further meeting, falling to 30.9% (95% CI: 26.6 - 35.5) in 2013. Linked to this, only a minority of families said they were told how the genetic counselling service could help them in the future; 28.9% in 2003 and 31.0% in 2013.

7: Signposting to peer support: Signposting of families by all specialists to some form of peer support group has risen significantly, from 34.8% of families in 2003 to 58.7% in 2013. Respondents were increasingly likely to be signposted to Unique (26% in 2003 rising to 67% in 2013). Only a small number of respondents stated that the genetic specialist tried to put them off contacting other affected families (7.3% in 2003 and 3.9% in 2013). Nevertheless, very few respondents (7% in both time periods) reported that they were offered any help to contact other RCD families.

3.4 Recommended improvements identified

Table 3 lists the recommended improvements identified by experts based on survey responses. The penultimate column identifies which are included in the NHS England service specification for organisations funded to provide specialised medical genetics services [20]. This indicates that 12 out of 28 recommendations identified by the present study are already included in service specifications. However, aspects which are missing include: a) education of non-clinical professionals; b) recommended speed of testing; c) six practical recommendations for communication of test results; d) need to indicate waiting time for referral to a genetics expert; e) five specific recommendations for conduct of consultation with genetics expert. For section f), all recommendations identified by the current study are included in the service specification. The final column shows levels achieved as reported by respondents for selected recommendations included in the service specification. These range from 36% to 80%.

4. DISCUSSION

The necessity for patient reported outcomes (PROs) in clinical genetics services has been identified in a recent review [7]. However, provision of services to families who have a child with a RCD is acknowledged to be exceptionally challenging [4-6]. There is also limited research evidence. A review of research into clinical genetics services and the patient perspective which identified 102 articles [21] found only one focused on these families [3]. The recommendations identified in the current study are novel because they are based on the real-life experiences of over one thousand families living with RCDs. As PROs become more important in performance management and funding of health services [7], sustained capture of the experiences of such families will be a key challenge [1]. Although a recommendation for 'sustained patient involvement in rare disease service provision' was embedded in the UK strategy for rare diseases, the overall strategy implementation plan for England has only recently been announced [22].

The large scale surveys reported here show that, although families' rating of service quality has improved over time, key aspects of the 'patient journey' have not and require improvement. Although agreement on key PROs for genetic services is generally acknowledged to be challenging for RCD cases [7 23], our surveys do highlight a number of simple improvements which might be easily introduced and which are indicated elsewhere. For example, a review of guidelines from 18 organisations in six countries on communication of genetic information to families concluded that there was a significant gap in terms of the professional's role in assisting clients to find options for continued support [24]. This is a key finding identified from our surveys. Our results also echo evidence from US research which found that parents of children with RCDs were largely disappointed in the counselling they received, although this was a small-scale study [3].

Although international guidelines for clinical genetics professionals largely cover the professional-client r**elationship**, including respect for the client, maintaining confidentiality, and enabling clients to make informed independent decisions [25 26], they do not include more practical PROs such as those reported in the present study. Other recommendations, such as those produced by Rare Disease UK (a project of the charity Genetic Alliance UK) mostly concentrate on higher level activities (e.g. commissioning and planning of services for rare diseases) with some general recommendations to improve information and support [27]. More recent recommendations for reporting the results of diagnostic genetic tests primarily focus on providing patients with information on how to manage their own condition, something which is less relevant for families of children with RCDs [28]. However, the most recent service specification for organisations providing specialised NHS medical genetics services does include some, but not all, of the recommendations identified in the present study [20].

Meanwhile, international evidence has emerged of large variations in clinical genetics practice, leading to an increased interest in defining the quality of services and improving delivery models [29-31]. Core competences and a code of practice have been produced for European health professionals [19 32], based on research by Skirton et al. [33] and approved by the European Society of Human Genetics [34]. To date such recommendations are based on the subjective views of professionals, rather than evidence-based, data on user experience. More recently**, t**he US National Society of Genetic Counselors launched a series of new *Evidence-Based Clinical Practice Guidelines* although up to now these do not include RCDs, only Fragile X Syndrome and Down Syndrome. (http://www.nsgc.org/practiceguidelines).

Service quality for RCD cases will inevitably be influenced by the availability of genetic specialist expertise. In this respect, the UK appears to be fortunate, with a higher number of genetic counsellors/nurses per million population than other European countries [31]. The existence of a long-established Rare Chromosome Disorder Support Group also differentiates the UK from other countries. As the rates of RCD diagnoses rise significantly, thanks to wider use of microarray analyses and the anticipated introduction of next generation DNA sequencing into routine clinical practice, combined with the fact that RCD cases are inevitable geographically widespread, the role of non-geneticist clinicians will inevitably continue at various stages of the patient journey, reinforcing the need for common guidelines, multidisciplinary teamwork, audit checklists, training and coordinated care pathways [35 36 37].

We recognise that the genetic and genomic testing and service landscape in the UK is also developing at a tremendous pace, not least because of the 100,000 Genomes project [38], the creation of 13 Genomic Medicine Centres across the UK [39], the Genomics England Clinical Interpretation Partnerships (GeCIP), designed to improve the accuracy and reliability of information fed back to patients [38]; and a drive by Health Education England to educate non-genetics healthcare and other professionals in genomic medicine [40]. It is therefore imperative that the value of the expertise of UK families affected by RCDs is not lost in the rapid pace of developments in genomics per se for identifying current and future needs. Although patients' and professionals' views may differ, there does appear to be a level of consensus on important domains such as: decision-making, knowledge of the genetic condition, perceived personal control, risk perception, diagnostic accuracy, and satisfaction/ quality of life [41]. Also, since clinical genetics services in the UK are currently delivered through a network of 23 centres, this network could facilitate the introduction of a coordinated strategy to support these families, although there is currently no designated centre of excellence specific to RCDs to take the lead. It is possible that an holistic RCD-specific service might be introduced by the newly-emerging rare disease centres, such as those in Birmingham [42] and London [43].

Our study inevitably has a number of limitations that should be borne in mind when considering the findings and subsequent recommendations. Firstly, some bias in responses is likely as participants were recruited from a specialist support group and therefore respondents may be different from other UK families with an RCD child. Secondly, it is possible that parents in Unique may be more knowledgeable because they are part of a well-established support group and have higher expectations (e.g. in terms of the information required) than people who do not belong to such an organisation. Finally, there may be recall inaccuracy since, in some instances, the survey requested information from families sometime after the event.

Conclusions & Recommendations:

These surveys of Unique members address the lack of data on genetic diagnosis and counselling care pathways experienced by families of children with RCDs. Recommendations are offered in the spirit of constructive collaboration to assist clinicians to best meet the needs of patients and their families [20]. The findings set baseline data for the experiences of families in 2003 and 2013. The intention is to repeat the surveys in 2018/2019 to gather patient-reported experiences as the new streamlined genetics service configuration is rolled out across the UK, and then again in 2021/2022 when new genetics services and implementation plans for the rare disease strategy are well embedded in the UK service provision. We consider that establishment of this form of longer term overview of user experience is particularly important, not least because diagnoses and genetic counselling are likely to be increasingly provided by non-geneticist clinicians.

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Legends:

Table 1: Questionnaire Content for Stages 1-7 in the Patient Journey

Table 2: Questionnaire responses and changes over time (2003 – 2013)

Table 3: Study Recommendations vs NHS England Service Specification, and Levels Achieved.

Figure 1: Rating of Genetic Counselling Services