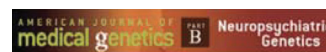


## ORIGINAL ARTICLE



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# Improving access to genetic testing for adults with intellectual disability: A literature review and lessons from a quality improvement project in East London

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## Abstract

Recent advances in genetic research have led to an increased focus on genetic causes of intellectual disability (ID) and have raised new questions about how and when clinicians offer genetic testing and the nature of communication around this decision with patients and carers. Determining the right approach to such discussions is complicated by complexities of communication, consent, and capacity and ethical concerns about genetic testing in this population. In this article, we briefly discuss the recent advances in genetic research relevant to people with intellectual disability, highlighting the challenges that might arise when undertaking genetic testing in this population. We then describe how we have used a Quality Improvement methodology to develop a clinical pathway for routine genetic testing for adults with intellectual disability in a clinical setting in East London.

## KEYWORDS

capacity, ethics, intellectual disability, neuropsychiatric genetics

## 1 | INTRODUCTION

People with an intellectual disability (ID) are defined as having significant impairments in their intellectual and adaptive functioning, with onset before the age of 18 years. Intellectual disability can be classed as mild, moderate, or severe depending on the extent to which it impacts on a person's life; the majority of affected individuals falling into the mild category. Intellectual disability affects approximately 2–3% of the general population (Daily, Ardinger, & Holmes, 2000) and over 1 million people are currently considered to have an intellectual disability in the United Kingdom (PHE, 2016). Intellectual disability is caused by a clinically heterogeneous spectrum of disorders, some of which are genetic, some environmental, though many have an unknown cause.

Recent advances in genetic research have led to an increased focus on genetic causes of intellectual disability and have raised new questions about how and when clinicians can or should offer genetic testing. Discussing genetic testing with patients and carers can be complex and is often avoided by clinicians who do not do it regularly, for many different reasons. Such discussions may be complicated by complexities of communication, consent, and capacity as well as ethical concerns about genetic testing in this population.

Clinical Genetics services are well established in the United Kingdom and are based in regional centers, which cover the United Kingdom, usually with a network of outreach clinics. Clinical geneticists provide a diagnostic service alongside expertise in explaining genetic results and genetic testing in a variety of situations, including presymptomatic testing for late onset genetic disorders, and prenatal testing. Clinical genetics teams would be able to see individuals or families in whom a genetic disorder or susceptibility had been identified, but do not have the capacity to see everyone undergoing genetic testing.

Katherine Adlington and James Smith contributed equally to the preparation of this manuscript and are considered joint first authors.

In this article, we briefly discuss the recent advances in genetic research relevant to people with intellectual disability, highlighting some of the challenges that might arise when offering and undertaking genetic testing in this population. We then describe how we have used a Quality Improvement (QI) methodology to develop a clinical pathway for routine genetic testing for adults with intellectual disability in a clinical setting in East London. This pathway mainstreams the approach, with the initial discussions and testing being performed by the ID team, and patients being referred to clinical genetics services if abnormal or uncertain results are obtained.

## 2 | LITERATURE REVIEW OF GENETIC TESTING FOR PEOPLE WITH INTELLECTUAL DISABILITY

### 2.1 | Advances in genetic research in intellectual disability

Recent advances in clinical genetic techniques and pathways have meant genetic testing now offers more diagnostic information than ever before.

Testing for major chromosomal abnormalities, for example, Fragile X has long been a recognized part of the diagnostic pathway for people with intellectual disabilities (Miller, Adam, Aradhya, & Biesecker, 2010). G-banded karyotyping has been the standard first line test for detecting these major chromosomal abnormalities for the past several decades. However, karyotyping for chromosome abnormalities has now been replaced by chromosomal microarray analysis, also called array comparative genomic hybridization (aCGH). aCGH also analyses chromosomes but at a much higher resolution than karyotyping. This improves the diagnostic yield but also detects more changes of unknown significance, which may require further family testing or input from clinical genetics services for interpretation. Microarray testing has also led to the discovery of a number of neurosusceptibility variants. These are very small chromosome changes, which increase the chance of an individual having a variety of problems, such as intellectual disability, autism, attention deficit hyperactivity disorder (ADHD), or seizures and may increase the risk of some psychiatric disorders in adult life. Neurosusceptibility variants can be found on a number of different chromosomes and may be deletions or duplications. Not only do the effects vary from individual to individual, but the penetrance also varies (Kirov et al., 2014). Some of the neurosusceptibility variants may affect the majority of people who have the change (high penetrance) but others may only affect around 10% of individuals with the change (low penetrance). This means that in a family, there may be affected and unaffected people with the same chromosome result. The effects are impossible to predict, making prenatal testing and testing of young children problematic, practically and ethically.

Recent research found that around 10% of adults with presumed idiopathic intellectual disability presenting to psychiatric services had likely neurosusceptibility variants, with deletions and duplications at 15q11-q13 and 16p11.2-p13.11 being most frequently observed (Wolfe et al., 2016). Other recent research has shown that patients

with schizophrenia and neurosusceptibility variants were significantly more likely to have lower IQs (Lowther et al., 2017).

The newer tests such as aCHG are identifying an increasing number of variants. Some of these are associated with a risk of intellectual disability, autism, or schizophrenia while the significance of others remains unknown. However, there is relatively little data about the clinical outcomes associated with identifying these new, complex genetic variants.

aCGH does not detect very small deletions or duplications or mutations in single genes so is not a "perfect or complete" genetic test. Many single genes are known to cause syndromic or nonsyndromic ID. Traditionally, specific gene testing was targeted at genes, which match the clinical phenotype, usually based on dysmorphic features, but this approach is likely to change when exome or genome testing is introduced. However, the specific dysmorphic, physical, or behavioral phenotypes may be nonspecific or subtle, so many single gene disorders have probably been undiagnosed clinically for many years, such as Weidemann Steiner Syndrome (Jones et al., 2012).

Over the last few years panel testing has been introduced and developed, which tests a number of genes known to be linked with a particular phenotype, such as Noonan Syndrome or early onset epilepsy. This has replaced the standard approach of sequentially testing different genes.

In turn, panel testing is now being replaced by whole exome (WES) or whole genome sequencing (WGS), though this testing is not yet routinely available in the public health system in the United Kingdom. Each individual has a number of genetic variations, most of which are not significant. The challenge in introducing WES or WGS is the interpretation of the findings—both in assessing the nature and significance of a particular variant and whether the gene involved is likely to be contributing to the phenotype (primary finding) or whether it may be significant for unrelated medical problems, such as susceptibility for cancer (secondary finding), which may need to be discussed with the individual and have implications for other family members.

WES and WGS will undoubtedly improve the diagnostic yield, particularly for nonsyndromic ID. However, the results will be more complex and close interaction between clinicians and laboratory scientists will be necessary for accurate interpretation of the results.

One Dutch study retrospectively examined Whole Exome Sequence data for 370 patients with ID finding a diagnostic yield of 35 and an 80% reduction in healthcare costs per patient after genetic testing regardless of diagnostic result (Vrijenhoek et al., 2018). However, studies using patient reported outcome measures are sparse. Gathering evidence about outcomes for patients following genetic testing, such as effect on quality of life or health status, might build the confidence of intellectual disability healthcare teams when both offering tests and reporting results back to patients.

### 2.2 | Current pathways for genetic testing in intellectual disability

There are existing local pathways through which clinicians can gain support to offer genetic testing to people with intellectual disability.

For example, in the United Kingdom, there are 25 regional genetics services, covering the whole of the country. These services provide a diagnostic service to adults and children, “genetic counseling” genetic testing when indicated and desired by the individual, for example, presymptomatic testing, carrier testing and prenatal testing and work closely with relevant teams, such as those providing preimplantation genetic testing.

Most centers request that microarray testing is initiated by the referring team at the time of referral to improve the efficiency of an appointment.

Karyotyping has largely been superseded by higher resolution assays (as discussed above). aCGH is recommended as the first-line genetic investigation for ID in the United Kingdom, United States, and many other countries (Moeschler & Shevell, 2014). This change in clinical practice has implications for all patients. Not only should patients be offered aCGH on first presentation to services but as aCGH gives a higher yield than karyotyping, aCGH is recommended even if karyotyping has been performed in the past. Fragile X testing should also be considered a routine test for individuals with ID (excluding males with microcephaly). In a minority of individuals, there are obvious features of a syndromic condition, and in those individuals, specific gene testing may be more appropriate as the first-line investigation. However, DNA from aCGH testing will be stored and can be used for specific testing at a later date, so further tests will not need new blood samples.

A recent survey of Child and Adolescent Psychiatrists and Intellectual Disability psychiatrists in the United Kingdom found these doctors thought there needed to be better training and closer links with regional genetics services and that they would prefer to refer a regional genetics service than order a genetic test themselves (Wolfe et al., 2018). However, genetics services do not have the capacity to see people just for testing and nongeneticists need to initiate the process, if a patient and family wish to explore genetic testing. Their main concerns when considering genetic testing for a patient included the lack of available treatment, implications for insurance, and issues around counseling. Interestingly, intellectual disability psychiatrists were much more likely than Child and Adolescent Mental Health consultants to have either ordered a genetic test or referred a person to clinical genetics services in the past year (90 vs. 68%).

There has also been increasing interest in collecting genetic data for a range of different rare disorders and cancers to try to identify etiological variants. In the United Kingdom, the national 100,000 Genome Project has performed whole genome sequencing on patients with certain clinically indicated conditions (rare conditions and cancers, including many neurodevelopmental disorders). Results will be fed back to patients but is not a part of “standard practice” local pathways. Due to increasing discoveries of risk-associated copy number variants (CNVs) in schizophrenia and anorexia nervosa, both of these conditions were added to the 100,000 Genomes Project as eligible conditions in January 2018 (Genomics England, 2018). This project closed for recruitment in September 2018 but it is hoped the experience gained will be used to help develop and roll out such testing into the NHS setting.

## 2.3 | Why is genetic testing important for people with intellectual disability?

Although this emerging evidence suggests that genetic causes may have a greater role in the development of intellectual disability than previously understood, there are other important reasons to prioritize genetic testing within this vulnerable group (Palmer et al., 2014; Thygesen et al., 2018).

The presence of an intellectual disability may result in diagnostic overshadowing whereby mental or physical illness may go unnoticed and attributed to the disability alone (Jopp & Keys, 2001). Combined with increased rates of physical and mental illness, this can lead to complex presentations and the use of psychotropic drugs to manage challenging behavior without a diagnosis of severe mental illness (Kerr, 2004; Sheehan et al., 2015).

It is therefore imperative to explore etiology of intellectual disability and challenging behavior and ensure awareness of behavioral phenotypes associated with a genetic disorder. For example, Fragile X is associated with attention deficit hyperactivity disorder (ADHD) and complex partial epilepsy (Kidd et al., 2014; Lozano, Rosero, & Hagerman, 2014). Self-injurious behaviors are more common in Cornelia de Lange, Lesch Nyhan, and Smith-Magenis syndromes and individuals with Prader-Willi may develop affective or psychotic illness (Soni et al., 2008).

In the Wolfe et al. (2018) study cited above, the psychiatrists were asked to estimate the percentage of people with intellectual disability for whom genetic factors make a significant contribution toward the cause of their intellectual disability. The mean estimate was 39.6% *SD*  $\pm 3.9\%$ , however estimates of percentage caseload with an established diagnosis were just 10%, highlighting a significant proportion of people who potentially have an undiagnosed genetic disorder. Many clinicians in the survey expressed concern that diagnosis would not change management. Yet, there are well-established benefits in screening for disorders such as 22q11.2 deletion syndrome (Habel et al., 2014) and Tuberous Sclerosis (Northrup et al., 2013), which highlight the importance of identification.

People with intellectual disability are known to have poorer access to healthcare and experience health inequalities, including premature death, as a result (Heslop et al., 2013; Iacono et al., 2013). Identification of genetic syndromes that cause the intellectual disability can be very helpful in addressing health inequalities, especially if known physical disorders are associated with the condition. Health inequalities could be addressed by having individualized care plans (Health Action Plans) addressing the need for health interventions and future screening (Department of Health, 2001), as well as other interventions like education of health professionals in making reasonable adjustments for people with intellectual disabilities, and the introduction of primary care and general hospital liaison nurses (Michael & Richardson, 2008; Walsh, Handley, & Hall, 2014).

There are not only proven management benefits but also potential benefits in terms of understanding a person's condition, both for the person with an intellectual disability and their family and carers. Having a clear diagnosis or biological explanation for their intellectual

disability might be useful for some people and their families. There is research establishing a benefit to mothers in receiving a diagnosis for a child with ID (Lingen et al., 2016), however, this evidence does not extend to explore the impact of a genetic diagnosis for adults with ID. It also might provide information about genetic risk within a family for the person with ID and their relatives, as discussed below.

## 2.4 | What are the specific challenges to genetic testing for people with intellectual disability?

For many adults with intellectual disabilities, the possibility of accessing genetic testing is only a recent development. Although there are many potential benefits, as described above, this development poses many concerns for professionals working with this population and their families. These concerns might include raising the possibility of genetic testing, undertaking the testing, and considering the potential implications.

A key issue is that of determining capacity to consent to genetic testing; and should an individual lack this capacity, determining whether undergoing the testing is in their best interests. Results of microarray analysis can be complex to interpret and communicate to patients or carers and an even greater challenge if the person has an intellectual disability. The complex nature of genetic testing can potentially complicate the issue, with many concepts often not fully understood by clinicians who have greatest contact with this client group (i.e., clinicians working in community intellectual disability services). Indeed, a study of genetic carrier genetic screening programs concluded not only that pretest and posttest counseling was of the utmost importance, but also that such counseling should be provided by a clinician with expertise in communicating genetic information (Cho, McGowan, Metcalfe, & Sharp, 2013). This highlights, therefore, a possible training need for clinicians in community intellectual disability services if they are to take a lead in offering genetic testing.

A further issue in relation to genetic testing in this population is the consideration of the role of family in providing support. For many individuals, their family members play a key role in their support network and may be their primary source of support. Family members need to potentially be involved in the process of genetic testing, as they may need to take a key role in communicating health information and supporting people in deciding whether to accept or decline testing. As with other genetic disorders, family members may have interests themselves in the result of genetic testing, potentially influencing the person with intellectual disabilities' decision making to take up testing.

Given the recent developments in genetic testing, there is a dearth of research into the psychosocial impact on adults with learning disabilities and their families regarding genetic testing. There is an increasing literature pertaining to prenatal genetic screening for conditions such as cystic fibrosis and genetic testing in adults for genetic disorders, such as Huntington's disease. (Axworthy, Brock, Bobrow, & Marteau, 1996; Broadstock, Michie, & Marteau, 2000; Ioannou, Massie, Collins, McClaren, & Delatycki, 2010). However, these situations are not comparable with diagnostic testing in individuals with ID

and it would not be appropriate to extrapolate their findings to this population. Clinical experience has shown that parents of children with genetic disorders often describe a sense of relief at knowing the cause of the child's problems and feeling that they can take action if they wish to avoid having another child with similar problems. However, again this is a different scenario to obtaining information on an adult with ID. It is likely to be more straightforward to assess the impact on family members rather than the affected individual.

Other research has demonstrated that significant social and cultural inequalities exist in knowledge about testing (Green, Hewison, Bekker, Bryant, & Cuckle, 2004). Given that social inequalities are often present in the ID population and people with ID come from all cultural and ethnic backgrounds there is a clear need to consider access to and accessibility of knowledge and information regarding genetic testing to ensure that this opportunity is genuinely available to all.

## 3 | LEARNING FROM A LOCAL QUALITY IMPROVEMENT PROJECT IN EAST LONDON: HOW CAN HEALTH CARE PROFESSIONALS OFFER GENETIC TESTING TO PEOPLE WITH INTELLECTUAL DISABILITY?

Given the identified challenges to offering genetic testing to people with intellectual disability, it is useful to consider practical examples of how services have systematically introduced the option for genetic testing to people that they support. We report on the process and early outcomes of an ongoing project to offer genetic testing to adult patients in a community intellectual disability service in the United Kingdom—the Tower Hamlets Community Learning Disability Service in East London National Health Service (NHS) Foundation Trust. The project uses a Quality Improvement (QI) methodology (Bennett & Provoost, 2015). Through our learning, we consider how teams might address staff training needs, referral pathways to clinical genetics services and communication tools to make genetic testing more accessible for people with intellectual disability. The project has not yet formally started to record and report clinical outcomes from genetic testing, such as the results of testing and the impact on clinical care. Instead, we present results and learning from the initial phase of offering genetic testing to people presenting to the service and present two anonymized case studies for consideration.

### 3.1 | Quality improvement project team, aims, and methodology

The multidisciplinary QI team comprised a consultant psychiatrist, consultant clinical psychologist, speech and language therapist and learning disability nurse alongside rotating junior doctors, psychologists, and student nursing staff. The team received input from a consultant clinical geneticist from the local area. The team was working in a Community Learning Disability Service (CLDS) in the borough of Tower Hamlets in London.

Tower Hamlets is an Inner London Borough with relatively high levels of deprivation compared to other parts of London (Tower Hamlets Council, 2015). The total population is estimated at 298,108 in 2016 and expected to reach 345,360 by 2025. In March 2017, 961 people in Tower Hamlets were registered with General Practitioners (GPs) as having a learning disability and 46% of these were of Asian background, mostly Bangladeshi. Of these, 882 people are registered with the Tower Hamlets CLDS (Tower Hamlets Health and Wellbeing Board, 2017). The CLDS is jointly funded by health and social services, and provides comprehensive initial multidisciplinary assessment for people with learning disability. It then aims to meet people's health and social care needs through specialist health interventions, supporting access to mainstream services (including for example, GP and hospital care, employment, and leisure services) and providing social care packages.

The QI methodology used was based on the model for improvement framework, developed by associates in process improvement, and the adopted method of the Institute for Healthcare Improvement. The model involves asking three basic questions: What is the team trying to accomplish? How will the team know that a change is an improvement? What change can the team make that will result in improvement? The model then recommends using Plan Do Study Act cycles for each change idea to implement a specific change and measure whether the change is having the desired impact. The team continues to implement further change ideas over a defined period of time while measuring the specified outcomes continuously. The team was supported by central QI team, who are embedded as a core function of the East London Foundation NHS Trust and provide support to clinical teams throughout the Trust to undertake QI projects locally, including a specially trained QI coach.

### 3.1.1 | Quality improvement aim: What is the team trying to accomplish?

We aimed to offer genetic testing to 100% of eligible people at the point of entry to the Community Learning Disability Service.

Previous local practice had been to refer certain, high-risk patients for genetic testing to the regional clinical genetics service. Through our process mapping and discussions with the regional clinical genetics service, it became clear that if we were to promote much more widespread uptake of genetic testing, then we would need to move the initial offer of testing and arranging the test to the Community Learning Disability Service. In conjunction with the genetics service, we developed a referral and care pathway (see Supplementary File 1) describing this, and also a screening tool (see Supplementary File 2) to ensure that patients with dysmorphic features or family history of ID were referred to the genetics service at the time of testing.

At entry, our comprehensive multidisciplinary assessment is to determine eligibility for the service (i.e., the presence of learning disability) and health and social care needs. As part of this assessment, we determine the nature of the disability, so this is a good time both to discuss previous investigations into the etiology of the disability, and also to explore whether the person or their family are interested in further investigation. The team works with people from the age of

17. Many people enter the service at this age, but others enter in adulthood. Although numbers were not collected, it was found that most people had not had a genetic work up in childhood. For those where genetic work up had occurred, often this was before latest techniques such as microarrays had become available.

It also became clear that the project would need to empower and enable all members of the team to offer and discuss genetic testing at the referral assessment because this is routinely undertaken by different multidisciplinary team members and not only medical staff.

We decided to focus initially on genetic testing in new referrals as etiology of the disability would always be discussed as part of the assessment.

### 3.1.2 | Quality improvement measurement: How will the team know that a change is an improvement?

Baseline measurement indicated that almost no patients were being offered genetic testing at the point of initial assessment despite assessment of behavioral phenotype being suggested as part of National Clinical Guidance for Challenging Behaviour (National Institute for Health and Care Excellence, 2015). The team amended report templates so that every new patient assessment included a section recording whether the person had been offered genetic testing on initial assessment. This data was reviewed on a two weekly basis to see what proportion of new referral assessments had included an offer of genetic testing. An improvement would be considered to be an increase in the number of new referrals being offered genetic testing.

### 3.1.3 | Quality improvement process: What change can the team make that will result in improvement?

#### *Process mapping*

Once it was decided, the Community Learning Disability Service would offer and arrange the genetic testing, initial process mapping was performed.

The team used a variety of techniques to investigate the low offer and uptake of genetic testing at the start of the project including one-to-one interviews with staff, group discussions, and liaising with GPs and the genetics lab. Reasons for the low uptake included:

- Staff ambivalence about testing. This sometimes derived from personal feelings relating to the staff member having a child or other relative with a learning disability, and whether they personally would want to access testing or not.
- New staff having a lack of knowledge of genetic testing.
- Some staff feeling that this was a "medical" matter and should not form part of the multidisciplinary entry assessment.
- Practical problems accessing the test at GP surgeries (availability of the right bottles, transporting the bottles to the genetics lab).
- Reluctance to have blood tests, and the poor accessibility of saliva sampling kits.
- Anxiety about explaining results—from GPs and multidisciplinary staff.

**TABLE 1** Identified barriers to genetic testing and proposed change ideas

| Barrier to genetic testing                                                                                                         | Change idea                                                                                                                                                                                  |
|------------------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Staff ambivalence<br>Lack of knowledge of new staff<br>Staff considering this was “medical matter”                                 | Further education sessions for staff<br>• Emphasis on discussion time and eliciting staff views<br>Develop more accessible written materials for example, frequently asked questions leaflet |
| Reluctance to have blood tests                                                                                                     | Desensitization program for blood testing<br>Identify and source saliva testing kits                                                                                                         |
| Anxiety about explaining results                                                                                                   | Medical staff in team (psychiatrists and junior doctors in training) will take a lead on this and offer support<br>Onward referral to clinical genetics agreed for complex cases             |
| Patient and carer uncertainty about genetic testing, for example what it involved, the implications for the patient and the family | Weekly team meetings enabling discussion of patient and carer feedback.<br>Used to develop education materials for staff and patients                                                        |
| Low numbers of blood tests carried out in GP surgeries                                                                             | Discussion with GP surgeries about process of performing and sending genetic tests<br>Identify and source saliva testing kits                                                                |

- Difficulty in identifying eligible patients.
- An unclear line of responsibility for performing the tests.
- Poor knowledge of when to involve the local genetics service.

Driver diagrams were then formulated to identify multiple change ideas, which were incorporated into Plan do study act (PDSA) cycles as shown in Table 1.

#### *Plan-do-study-act cycles*

The team undertook five initial PDSA cycles based on the above change ideas. Further information about each cycle is below. Each cycle took about 2–4 weeks to implement and the team continued to measure the number of genetic tests offered every 2 weeks to determine if the cycle had made any impact. Sometimes this was difficult to judge given the small numbers of referrals overall.

#### **Box 1 (PDSA cycles)**

- Cycle 1: Devising and considering implementing a screening tool  
 Cycle 2: Developing educational material in easy read and distributing to staff, clients, and carers  
 Cycle 3: Use of an easy read consent form  
 Cycle 4: Training staff to offer genetic tests (online and face-to-face)  
 Cycle 5: Weekly discussion of genetic testing of new referrals at team meetings

family history, and dysmorphic features. However, after further discussion with our clinical geneticists, we decided to offer genetic testing to all people with learning disability entering the service as the learning disability itself was enough to warrant microarray screening. The screening tool was still helpful, however, to help identify relevant clinical information for the testing and whether to refer on to clinical genetics.

*Accessible written materials including consent form.* The team's speech and language therapist oversaw the development of easy read information sheets, poster, and consent forms (see Supplementary Files 3–5). We actively sought carer and patient input to help the development of these resources, which was invaluable.

*Education sessions.* Face to face and online staff training aimed to ensure that all members of the service, including psychologists, nurses, occupational therapists, speech and language therapists, and social workers felt enabled and empowered to offer genetic testing and seek support from medical staff when necessary. We provided three sessions initially, and provide ongoing ones to address limited uptake and staff turnover.

*Weekly clinical team meetings.* Previous research has identified varying knowledge on how to offer genetic testing in the intellectual disability population (Wolfe et al., 2018) and limited awareness of the impact this may have on families and carers (Lingen et al., 2016).

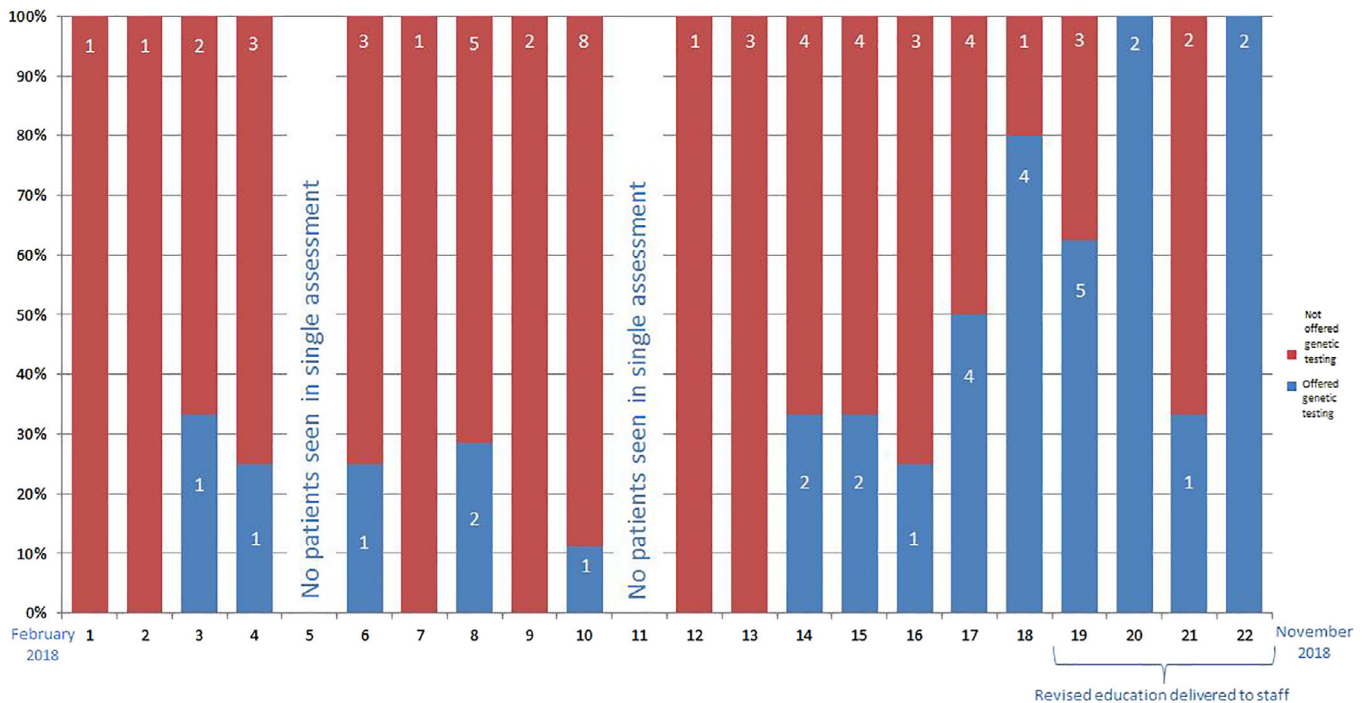
Scheduling regular discussion slots about genetics in weekly clinical team meetings enabled staff to ask questions and receive support around the decision making and genetic test offering process.

Regular discussion of patient and carer feedback at these meetings allowed development of awareness of the immediate impact of discussion and results. Patient feedback was collected regularly while designing and implementing the materials, which shaped the services ability to deal with sensitive issues such as reproductive decisions and risk of recurrence.

*Communication with local GP surgeries.* We found that even after agreement from the patient and/or family (as appropriate) had been obtained, a high proportion of people were not getting tested. One of

#### *Identified change ideas*

*Screening tool.* We developed a screening tool (see Supplementary File 2) to help people identify when a genetic cause for the learning disability was more likely, using criteria such as severity of learning disability,



**FIGURE 1** Proportion of patients offered genetic testing at initial assessment for the community learning disability service (February 2018 to November 2018). Each number on the x axis represents a 2 week period. That is, 1 = week 2; 2 = week 4, and so forth [Color figure can be viewed at [wileyonlinelibrary.com](http://wileyonlinelibrary.com)]

the major obstacles was identified as the separation between offering blood tests and providing blood tests. The offer is made by the CLDS, but as is common in community services the blood test itself is usually arranged through the GP surgery. We obtained feedback from GPs and identified a number of issues and solutions. Some GPs were anxious about explaining results to patients and families, but we reassured them that CLDS would do this, and refer to clinical genetics as indicated. Others were not sure how to transport the samples to the correct lab, which we reassured them the local hospital would do. We are also exploring the option of saliva sampling kits, which could be performed within CLDS.

### 3.2 | Quality improvement project results

Figure 1 shows the number of patients referred to the service by 2 week period and the number of patients offered genetic testing at this initial referral. Data were collected over 44 weeks. Referral numbers are relatively small. However, over the course of the 44 weeks of the QI project, the percentage of new patients offered genetic testing increased from an average of 14.5% in the first 8 weeks of the project to an average of 74% in the final 8 weeks of the project.

Figure 2 represents the same data in a p-chart—the graphical representation of data specifically used for QI projects. This shows data collected up to week 60 of the QI project. This graph shows that there was a meaningful increase in the average number of new patients being offered genetic testing after Week 30 according to QI methodology (labeled CL 21.74). These figures suggest the average number of new patients offered genetic testing increased from 20 to 60%.

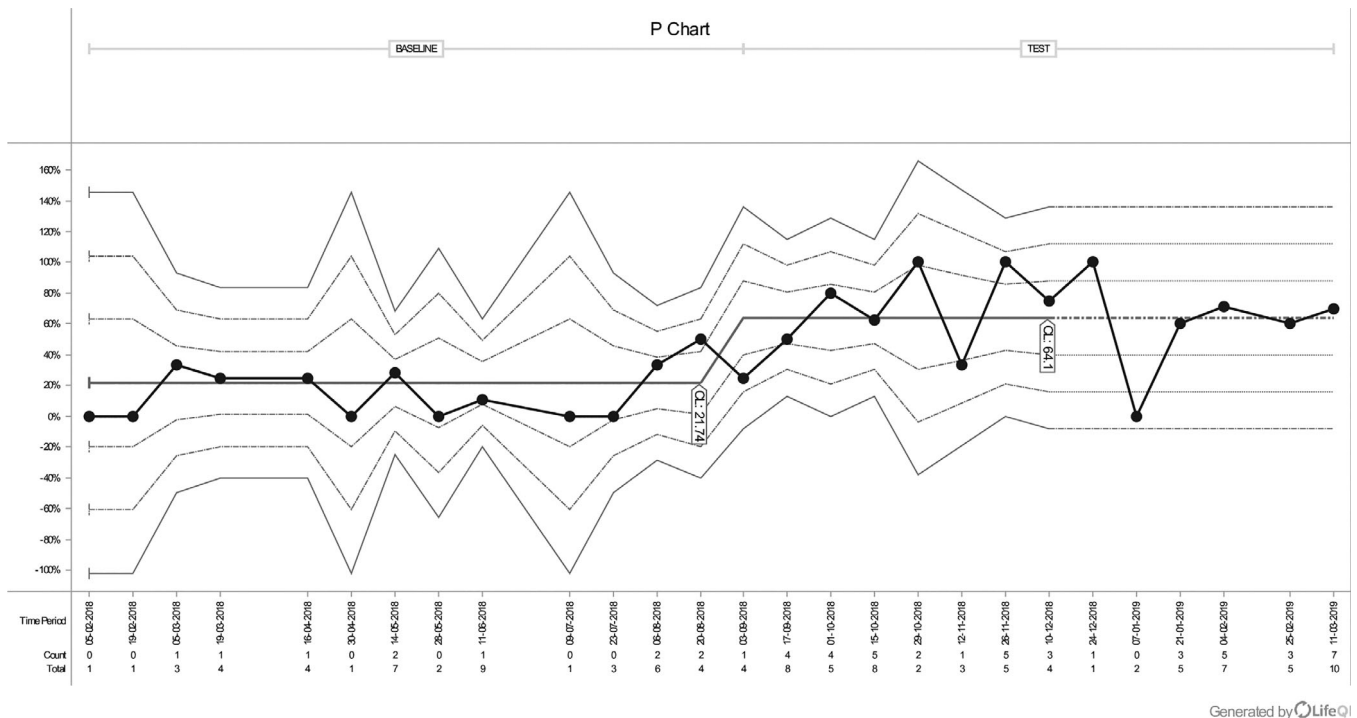
Although overall the team has achieved the aim of increased offer of genetic testing to newly referred people, the numbers are small in comparison to the total number of people registered with the CLDS, and working out how to offer testing to existing patients will be the next phase of the project.

Importantly, there is currently also a very low conversion from offer of test to taking the blood test in primary care, but now we have overcome barriers in primary care, and are obtaining saliva testing kits we are hopeful to address this.

### 3.3 | Reflections from this quality improvement project

The key lessons learnt from this project to increase the offer of genetic testing in a community learning disability service include:

- the importance of multidisciplinary involvement in developing pathways and offering genetic testing
- the importance of ongoing education for the MDT, including providing space for questions and concerns
- the importance of gathering patient and carer feedback in identifying potentially sensitive and complex issues, such as recurrence risk and family dynamics, and dealing with these responsibly
- the importance of clear easy read information in enabling patients to make the decisions around genetic testing
- the importance of engaging all stakeholders (e.g., GPs, patients, families) when improving genetic testing services



Generated by LifeQI

**FIGURE 2** Quality improvement project P-chart showing the proportion of patients offered genetic testing at initial assessment for the community learning disability service (February 2018 to March 2019). NB. "Count" on the x-axis represents the number of genetic tests offered and "Total" represents the total number of new assessments performed

- the importance of a streamlined and easy process of offering and arranging testing

Interestingly, while we might have anticipated patient factors being one of the major barriers to arranging genetic testing, clinician factors seemed to be more relevant in this project. In particular, lack of knowledge and confidence among team members proved to be a barrier to offering genetic testing in the service and there appears to be a clinician-related barrier in primary care, that is, preventing testing taking place.

Case study 1 and 2 were selected to highlight frequently observed themes in testing and reporting results following implementation of genetic testing.

### 3.4 | Text box: Case studies

#### 3.4.1 | Case study 1

Mr. A had a mild intellectual disability and was known to the Community Learning Disability Service because he attended one of the local day centers and also had a paranoid psychotic illness that had required inpatient admission and for which he was now taking treatment in the community. He lived in his own flat, with minimal support. He was noted to have microcephaly, slightly dysmorphic features, and a high-pitched voice. He was offered genetic testing to investigate potential causes for his disability, and he agreed to this. Testing showed that he had a 3q29 microdeletion. When we fed back the result, A was interested to know that this was the cause for his learning disability and his small head, and we explained it might be passed on through his family. His mother lived

abroad, but he asked if we could meet with her to tell her when she was next over. When she visited, we arranged two appointments to see her, but she canceled both appointments without giving a reason.

#### 3.4.2 | Case study 2

Mr. B had three young adult children. Both his sons had autism, one with a moderate intellectual disability and bipolar affective disorder, and the other with a severe intellectual impairment, autism, and hypomania. His daughter was unaffected. Both his sons were under the care of the Community Learning Disability Team, and had not had the cause of their learning disability previously investigated. It was explained to Mr. B that it was likely that there was a genetic cause that might explain his sons' autism and intellectual disability, and that it was easy to test for this, and might help us support his sons in the most effective way in terms of helping manage their behavior and optimizing their physical health. In addition, knowing the cause of their problems may give the family information about the chances of his daughter's future children having similar problems. However, Mr. B did not want to pursue testing, as he said their disability was God's will and it would not take their disability away. Neither of the sons had capacity to consent to the testing, so the team had to make a decision about what was in the sons' best interests, taking into account the father's view. The team was aware of the strong possibility of an X-linked inherited condition being the cause of the intellectual disability in the males of the family, and as a result has potential significance for their sister and her future children. The case highlights the difficulty balancing the competing rights of the family members involved and the difficulties parents have to input into making best interest decisions for patients who lack capacity to consent. The team

decided not to pursue testing for the time being, but to continue to discuss with the family, and perhaps involve the clinical genetic services in helping to make the final best interests decision.

## 4 | CONCLUSION

Recent advances in gene sequencing as well as the discovery of increasing numbers of pathogenic CNVs means that genetic testing can provide much more information for people with intellectual disability than ever before. Genetic testing may provide an explanation for an individual's intellectual disability and a diagnosis may lead to improved care and health outcomes, by allowing better prediction and management of associated behavioral, psychiatric, or physical health problems. It also enables prediction of recurrence risks for the offspring of the affected individual and their relatives and allows testing around a pregnancy, if that is chosen by an affected individual or family member. The process of arranging genetic testing or referring people to clinical genetics services is not new and is something that patients are entitled to, however, evidence suggests clinicians may be apprehensive about the processes and implications of genetic testing.

The real challenge is ensuring that this testing is as accessible as possible physically by designing appropriate testing and referral pathways. The first step to providing accessible genetic testing to people with intellectual disability is to ensure it is offered in an accessible and systematic way. We have reported on a QI project that has developed means to offer genetic testing to all new patients by a multidisciplinary team. We have worked with our local Clinical Genetics Service to develop a systematic, patient-centered way to offer accessible genetic testing to our patients, and overcome barriers to implementation.

Next steps for the project will be to refine our approach to communicating complex results in an accessible, meaningful way for our patients and their families. This will include collecting information on patient and carer experience and quality of life, accessibility, staff experience, and patient health outcomes such as changes in health status or changes to management and medication. We will continue to evaluate the process, including remaining open to potential deleterious effects of genetic tests, but believe that the processes can be rolled out to other units to benefit more patients and families.

## CONFLICT OF INTEREST

All authors have no relevant interest to declare.

## AUTHOR CONTRIBUTIONS

All authors contributed equally to the research and writing of this article.

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## SUPPORTING INFORMATION

Additional supporting information may be found online in the Supporting Information section at the end of this article.

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