Auditing the Psychiatric Caseload for Eligibility and Uptake of Genetic Testing

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Introduction

We are a multi-disciplinary service offering specialist healthcare and social support for those living with a learning disability (LD) and their families in the London borough of Tower Hamlets. Recent advances in genetic screening have allowed a better understanding of the genetic causes of learning disability. As part of a quality improvement initiative started in 2017, we have been offering suitable clients the option of genetic testing at the point of entry to the service. To further widen the availability of this testing, we have audited the current psychiatric caseload of patients who may have joined the service before genetic testing was routinely offered at initial assessment.

Aims

- Expand the current quality improvement project on offering genetic testing to include as many suitable clients in our service as possible
- Offer genetic testing to all eligible clients open to psychiatry
- Understand the demographics, potential aetiology of LD, and patterns of testing uptake in this population

Methods

We used information gathered at the initial assessment on entry to the service to identify those eligible for genetic testing. Those with a previously identified cause of their LD were not offered further testing. Suitable clients were offered blood testing at local hospital phlebotomy services and saliva testing either at home or at CLDS offices. We used descriptive statistics to analyse the findings.

Demographics

Our sample consisted of 154 patients between the ages 18-73 (Chart 1). Males outnumber females (98 versus 56). 56% are Bangladeshi, 23% white, and 6% Black which somewhat correlates with the general demographics of Tower Hamlets which is 32% Bangladeshi[1]. The majority of the patients have mild learning disability (Chart 2).

Epilepsy is overrepresented in this group (28, 18.2% versus national average of 0.5-1%[2]) as is hypothyroidism (11, 7.1% versus 1-2% nationally[3]) and Type 2 diabetes (17, 11% versus 6% nationally[4]).

Chart 1: Age distribution

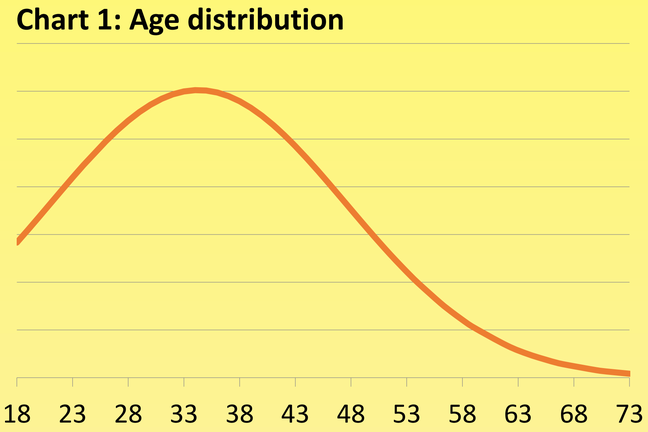
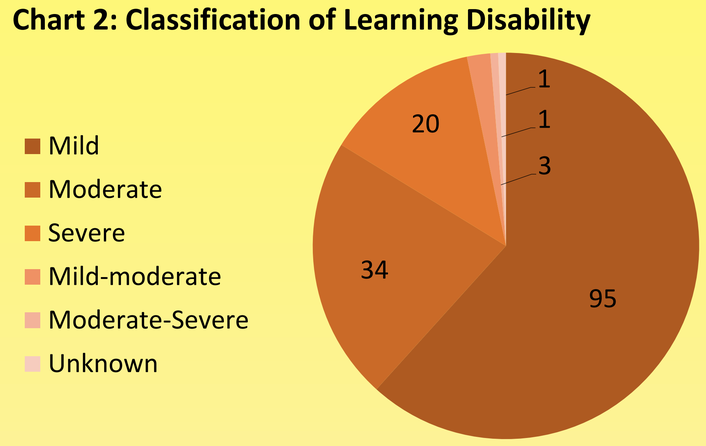


Chart 2: Classification of Learning Disability



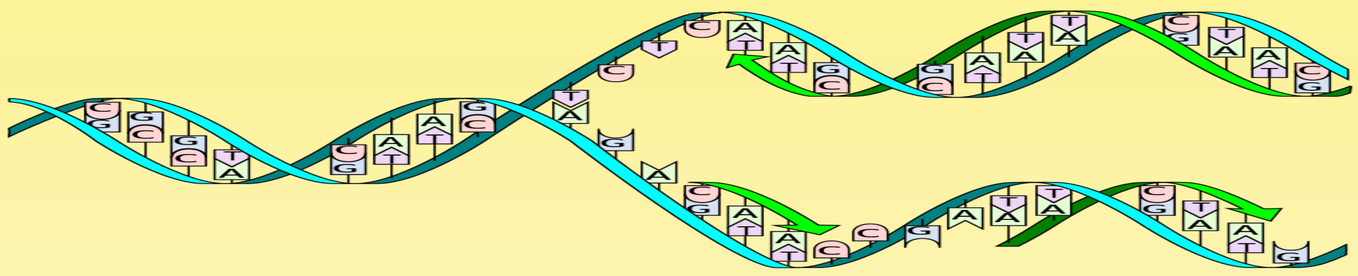


Chart 3: Uptake of genetic testing

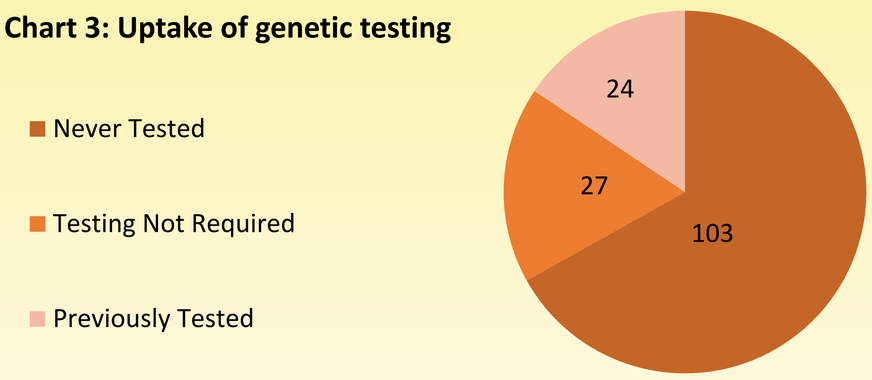
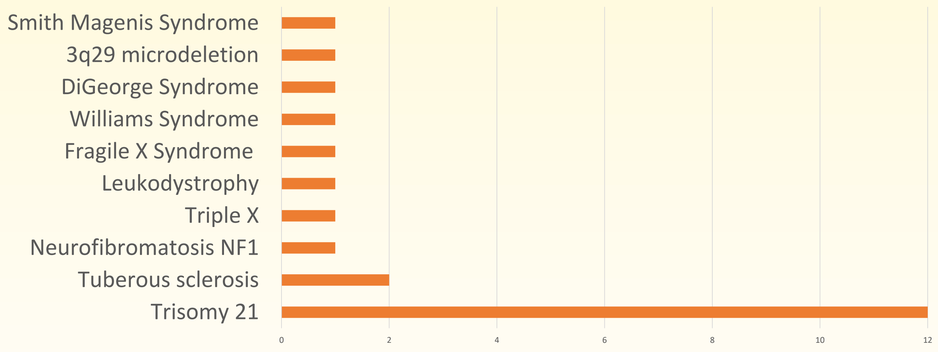


Chart 4: Previous genetic diagnoses



Cause of Learning Disability	Number of patients
Unknown	107
Perinatal	16
Trisomy 21	12
Other genetic condition	10
Head Injury	3
Neurodevelopmental condition	3

Results

Of the 154 audited patients open to the CLDS psychiatry team, 103 were eligible for further genetic testing, 27 were not eligible – mostly due to a clear, non-genetic cause of their learning disability- and 24 had already undergone testing (Chart 3). Of the 24 previous patients tested, 5 (20.8%) had no significant findings.

Of those with an established genetic cause, 12 (7.8%) had Trisomy 21, 2 (1.3%) had tuberous sclerosis, and there were 8 other unique genetic causes (refer to Chart 4). Established, non-genetic causes were less clear as this often relied on accurate recall of birth and developmental history. These included perinatal difficulties (16 patients), post-natal head injury (3), congenital and childhood infections (3), and neurodevelopmental conditions (3), such as agenesis of the corpus callosum and frontal lobe atrophy (refer to table).

There was no significant difference between the mild, moderate, and severe groups for eligibility for future genetic testing (p=0.973), previous testing (p=0.911), or those not eligible (p=0.987).

Discussion

Our aim for this audit has been to identify those eligible for genetic screening tests and to widen participation as part of a broader quality improvement initiative. Roughly two thirds of the psychiatric caseload is eligible for genetic testing but, for the majority, this has never been offered. Increasing access to testing has been facilitated by a new, comprehensive tracking system and staff training. We have begun contacting these patients and their families over the phone and in person at time of review.

For those that have declined testing, aversion to blood tests and a feeling that the result would not be helpful are the most common reasons. To overcome this, we offer saliva testing in the patient's home and we have redesigned our easy-read information leaflets to better demonstrate the benefits of testing with additional versions in Bengali. We hope this will better engage our patients and drive increased uptake of screening tests.

Furthermore, we predict a high diagnostic yield from genetic screening in our cohort. 79.2% of the patients already tested have yielded a positive result. Just over a third of our cohort have a diagnosis of autism, a condition in which approximately 25% of cases have a demonstrable genetic cause[5]. The presence of other mental health conditions further increases the likelihood of a genetic cause. In addition, there is an estimated 8% of children born to consanguineous marriages in Tower Hamlets which is linked to an increased susceptibility of inherited disease[6]. In 2014-2015, 18-25% of children seen with developmental delay in Tower Hamlets came from consanguineous parents and 80% of these speak Bengali or Sylheti at home[7].

References:

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