UNDERSTANDING THE VARIATIONS AND BARRIERS IN INVESTIGATION OF EARLY DEVELOPMENTAL IMPAIRMENT

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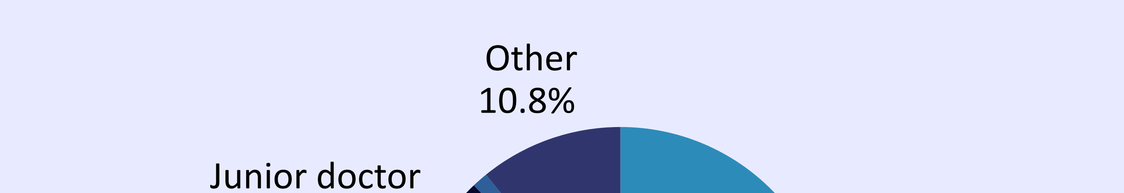
WHY DID YOU DO THIS WORK?

We aimed to understand the variation in practice and investigations, including genetic testing, for children with Early Developmental Impairment (EDI) in the East of England (EoE) and East Midlands (EM). We sought to identify barriers and propose potential solutions to standardise and improve these processes across the regions.

WHAT DID YOU DO?

An initial scoping survey across EoE was followed by a systematic review of articles from 2013 to 2023 towards formulating a proposed regional EDI investigation strategy. Working with stakeholders (Community Paediatricians and East Genomics) to establish its content, we distributed an electronic survey exploring attitudes around the current practices to paediatricians across EoE and EM.

Figure 1: 69% of clinicians performed genetic tests as a first-line investigation



WHAT DID YOU FIND?

Across EoE, 9 survey respondents identified 4 guidelines in use with differing recommendations on testing in EDI. Three centres reported following no guideline. The literature review explored recommended investigations for EDI, revealing possible publication bias towards genetic testing, being the focus of 16 of the 27 reviewed articles.We identified the highest overall diagnostic yield in whole exome and genome sequencing.

Figure 2: Roles of healthcare professionals responded to survey

Qualitative data from the survey (83 respondents) indicated that 69% of clinicians performed genetic tests as a first-line investigation (Figure 1), most commonly microarray (85%) (Figure 3).

Significant barriers were identified:

- 84% of respondents found it difficult to discuss genetic results with patients
- logistical issues such as sample collection and time for patient consent were highlighted

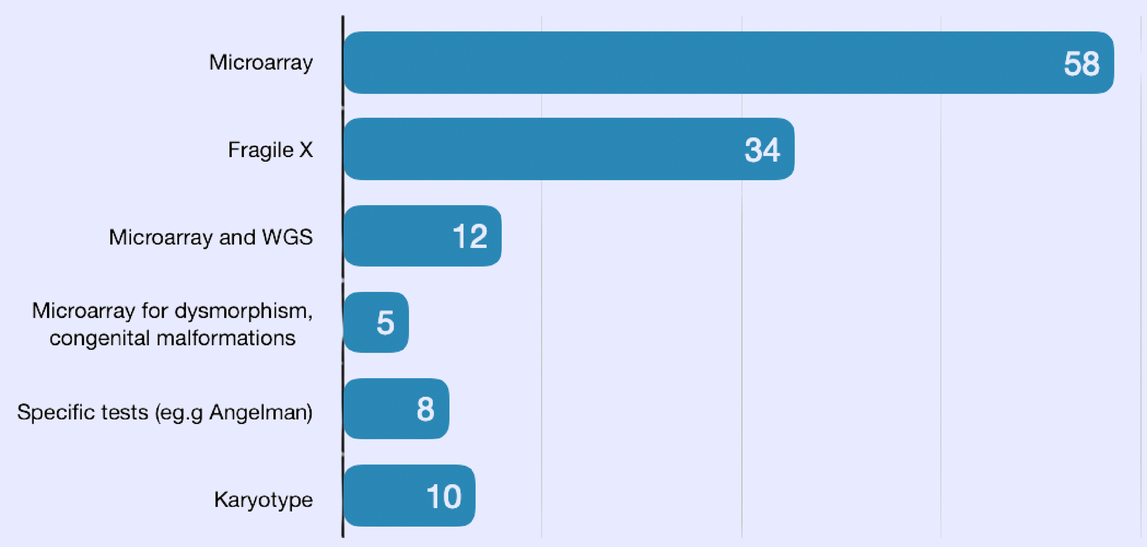


Figure 3: Commonly preferred genetic tests

Potential solutions identified by respondents;

- development of a regional EDI investigation guideline (92%)
- access to genomic practitioners to assist with consent and logistics (73%)
- establishment of regional neurodevelopmental genetics multidisciplinary teams (51%)
- implementation of electronic test ordering systems (63%). (Figure 4).

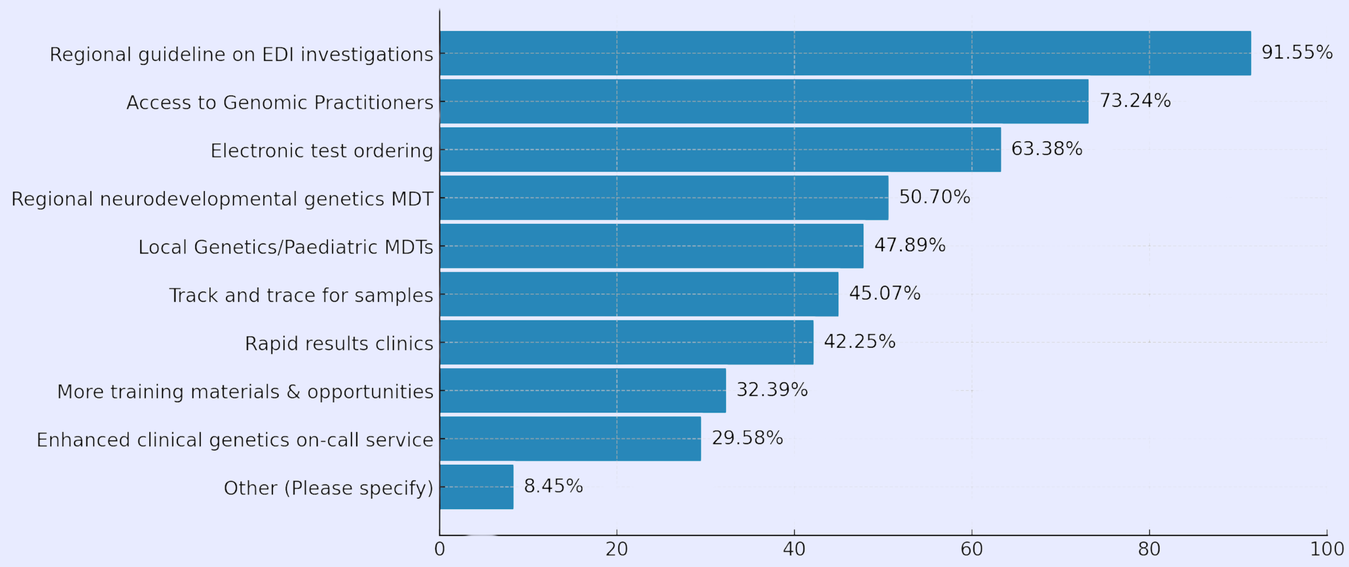


Figure 4: Suggested EDI investigation strategies

Significant variation in current practices;

- The majority relied on clinical acumen or informal assessments (89%), with only 13% using formal tools

Documentation of EDI severity was inconsistent;

- only 23% reported always documenting severity - methods varying between giving overall ratings (36%) and domain-specific assessments (49%).

97% expressed desire for a regional framework.

WHAT DOES IT MEAN?

Our findings highlight the challenges in overcoming variation in investigative approaches to EDI in particular relating to genetic testing. The data underlines the need for regional standardisation, increased education and support systems for clinicians. To address these issues, we are collaborating with paediatricians and geneticists to develop a practical framework that will facilitate consistent and equitable investigations across the region served by East Genomics.

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For questions and comments;
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