# Addressing Disparities in Paediatric Genetic Testing

## A Systematic Review of Engagement with Underserved Communities and Barriers to Access

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'Underserved populations' as per the NIHR definition are groups that are less well-represented in research than would be expected or desirable from population prevalence and healthcare burden.

## Background & Aims

Genomic testing is a key driver of precision medicine. Despite increasing uptake of genomic testing, there remains a problem of inequitable access particularly in underserved communities.[1] To understand the issues of disparity in access, we performed two systematic literature reviews with the following aims,

1. To evaluate the extent of genomic testing engagement in underserved communities within community /outpatient paediatric settings.
2. To identify barriers and facilitators to engagement with genomic testing in underserved communities.

## Methods

- Two systematic literature reviews
- Keywords relating to 'paediatric genetics', 'community settings' and 'underserved populations'
- Ovid, Medline, Embase, CINAHL and PubMed
- Screened by two independent reviewers and conflicts resolved by a third reviewer
- PRISMA approach and qualitative analysis (**Figure 1**)

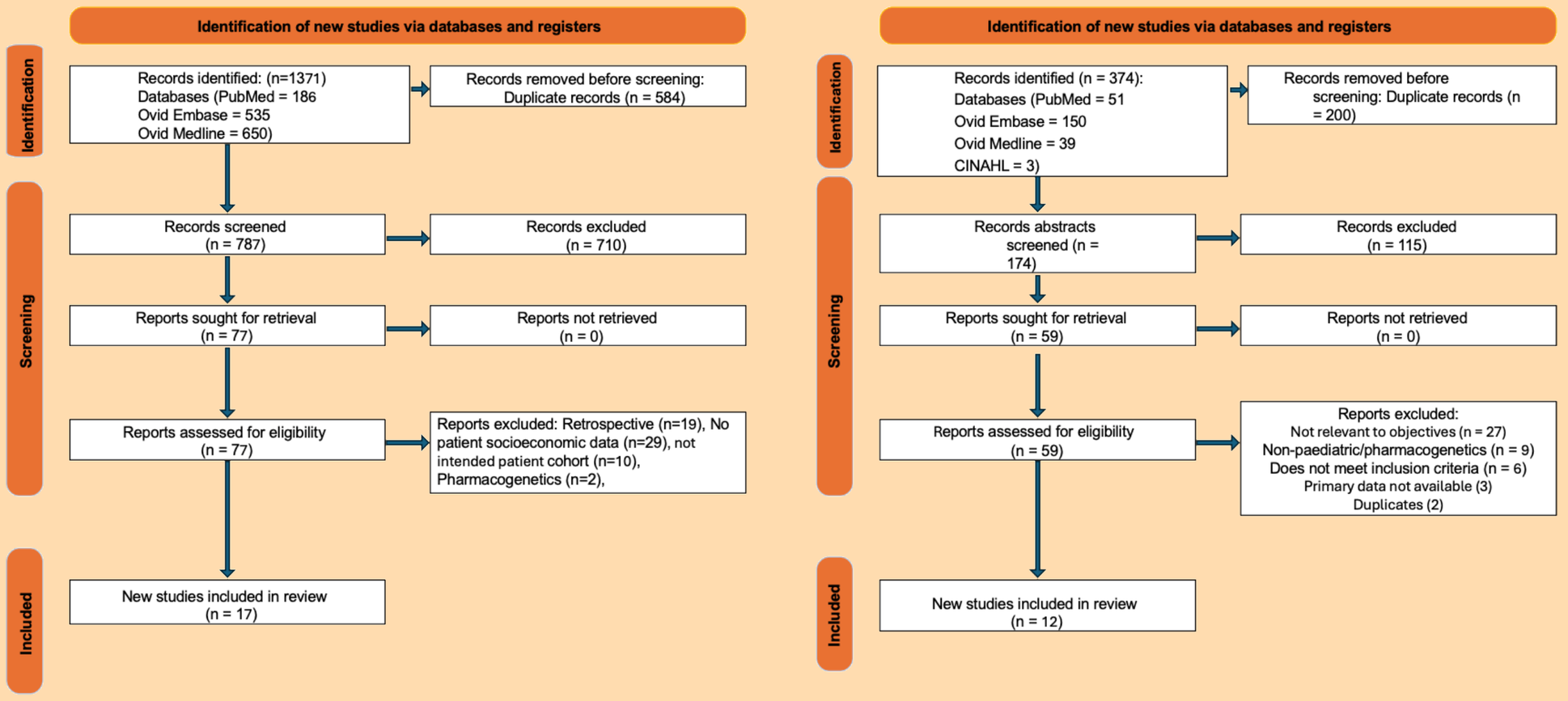


***Figure 1 PRISMA flowcharts for the systematic reviews undertaken. A:*** *Records screened for the evaluation of the extent of genomic testing reported in community or outpatient Paediatric settings.* ***B:*** *Records screened in the evaluation of barriers and facilitators for genomic testing in underserved communities.*

## Results

- Aim 1: 17 articles met the inclusion criteria. Our results show sparse reporting of cohort characteristics in the existing literature (**Table 1**).
- Aim 2: 12 articles met the inclusion criteria. Barriers and facilitators from our analysis are shown in **Figures 2** and **3.**

| Author | Year | Country | Ethnicity | SES | Education | Location |
|---|---|---|---|---|---|---|
| Symonds et al. | 2021 | Scotland | | | | |
| Downie et al. | 2019 | Australia | | | | |
| Forbes et al. | 2021 | USA | | | | |
| Harding et al. | 2022 | UK | | | | |
| Kim et al. | 2023 | South Korea | | | | |
| Zanoni et al. | 2023 | Switzerland | | | | |
| Ibrahim et al. | 2023 | Egypt | | | | |
| Durand et al. | 2023 | USA | | | | |
| Sebastin et al. | 2023 | USA | | | | |
| Chan et al. | 2021 | UK | | | | |
| Brozou et al. | 2018 | Germany | | | | |
| Yanes et al. | 2023 | Australia | | | | |
| Ververi et al. | 2023 | UK, Switzerland, Norway | | | | |
| Costain et al. | 2020 | Canada | | | | |
| Hou et al. | 2023 | China | | | | |
| Paola et al. | 2024 | Italy | | | | |
| Knight et al. | 2019 | USA | | | | |

***Table 1: Characteristics included in 17 articles used for the evaluation of the extent of genomic testing in underserved communities.*** *Green: adequate reporting, Yellow: inadequate reporting, Red: no reporting, Grey: not applicable*

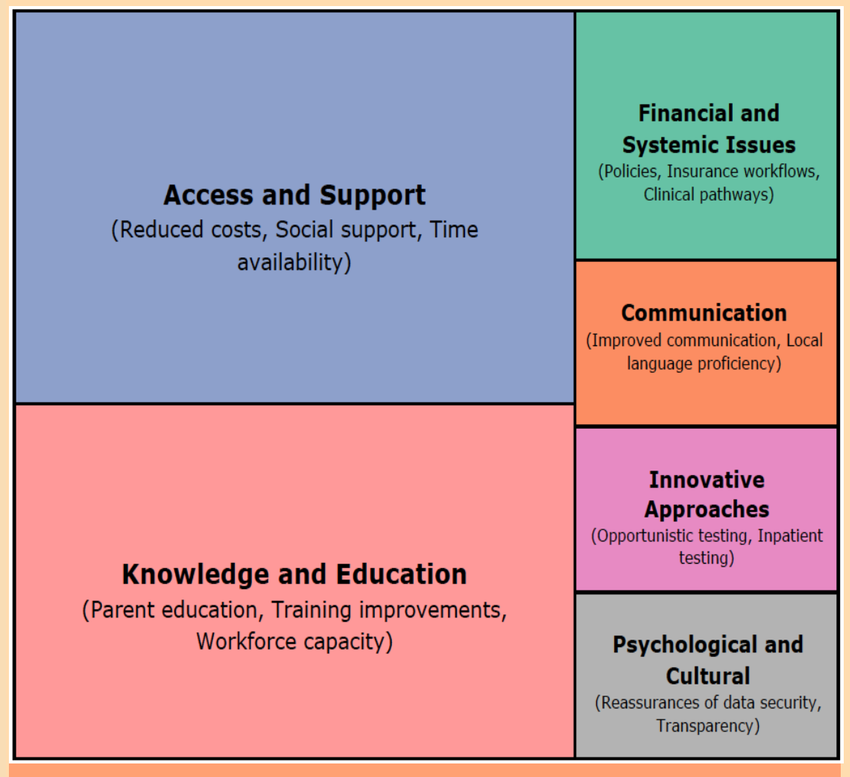


***Figure 2: Thematic analysis of codes generated across 12 papers relating to facilitators for engagement of underserved communities..***

## Conclusion

- There is limited consistent reporting on underserved population characteristics in paediatric genetic studies.
- We found a paucity of data on engagement of underserved communities within rare disease genetic studies (**Table 1)**.
- Key identified themes within the second review regarding barriers to engagement included lack of financial resources, access to genetic testing services, logistics in attending appointments, geographic isolation, language, socio-cultural beliefs, data privacy and ethical concerns. Facilitators followed similar themes (**Figure 2**).
- Solutions to increasing uptake could include strategies as highlighted (**Figure 3**).

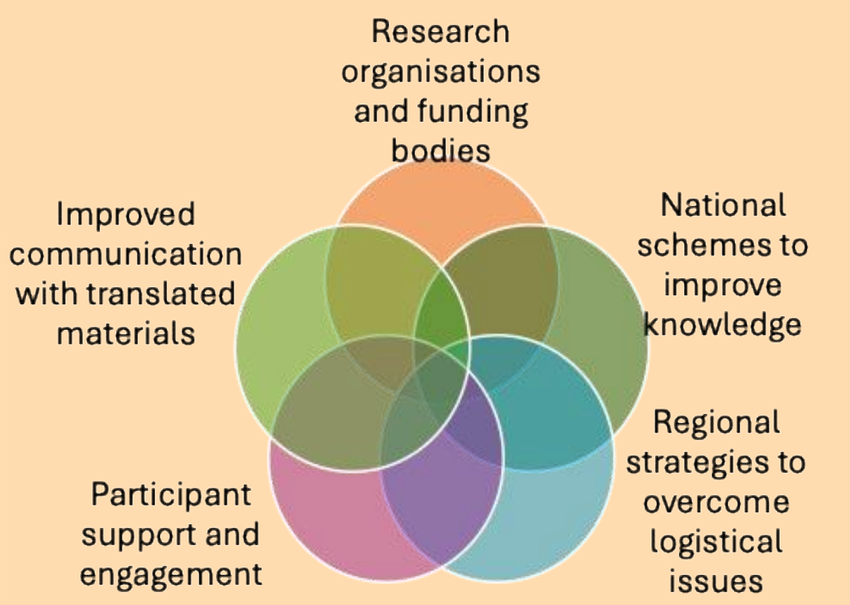


***Figure 3: Meaningful steps to improving uptake***

## Affiliations and Acknowledgements

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Reference: Bains, M., Qureshi, N., Bajwa, R. K., Leonardi-Bee, J., Hassanein, Z. M., Hassan, S., Jayes, L., & Bogdanovica, I Ethnic Inequities in Genomics and Precision Medicine. University of Nottingham. Retrieved from [https://www.nhsrho.org/research/ethnic-inequities-in-genomics-and-precision-medicine-review-report/]