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Systematic Review

Keywords: intellectual disability, genetic evaluation, barriers and facilitators, access to health care, qualitative evidence synthesis, epistemic justice

Posted Date: August 7th, 2026

DOI: https://doi.org/10.21203/rs.3.rs-10617927/v1

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Additional Declarations: The authors declare no competing interests.

Psychosocial and Cultural Barriers and Facilitators to the Genetic Evaluation Pathway in Intellectual Disability

A Qualitative Evidence Synthesis

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Abstract

Intellectual disability (ID) frequently warrants genetic evaluation, and international guidelines strongly recommend exome or genome sequencing as a first- or second-tier test in children with congenital anomalies or ID. Yet families often navigate a prolonged diagnostic pathway, and the psychosocial and cultural factors that shape actual access remain fragmented across the literature. We synthesised qualitative, mixed-methods and quantitative evidence on the structural, sociocultural and psychosocial barriers and facilitators influencing access to and engagement with the genetic-evaluation pathway among individuals with ID, their families and caregivers. Following PRISMA 2020 and structured with the SPIDER framework, we searched PubMed and Scopus for empirical studies published between January 2020 and March 2026. Qualitative and mixed-methods studies were synthesised using thematic synthesis; quality was appraised with the MMAT 2018 and confidence in each finding with GRADE-CERQual, while quantitative studies of access and uptake were retained as context. Fourteen qualitative and mixed-methods studies were included and six quantitative studies retained as context, spanning Europe, Australia, the United States, Israel, Japan and Brazil. Documented barriers outnumbered facilitators; this asymmetry is interpreted partly as a feature of the searched literature rather than an established property of the field. Higher-confidence findings concerned the exclusion of people with ID from decisions about their own care and the psychosocial burden on caregivers, whereas the fewer facilitators — co-designed accessible information, clinician toolkits and integrated care models — were supported with lower confidence. Access is shaped by human, social and structural factors alongside technical ones, with confidence graded throughout.

Keywords: intellectual disability; genetic evaluation; barriers and facilitators; access to health care; qualitative evidence synthesis; epistemic justice.

1. Introduction

Intellectual disability (ID) is a neurodevelopmental condition, originating during the developmental period, defined by significant limitations in intellectual functioning and in adaptive behaviour across conceptual, social and practical domains[1–3]. Given its frequently genetic aetiology, a genetic evaluation is often indicated, integrating family history, physical examination

and genetic counselling with molecular diagnostics — chromosomal microarray, whole-exome sequencing and whole-genome sequencing — to identify causal variants. International guidance reflects this indication: the American College of Medical Genetics and Genomics issues a strong recommendation for exome or genome sequencing as a first- or second-tier test in children with congenital anomalies or intellectual disability[4].

Despite the well-documented molecular yield of these technologies, the pathway from clinical suspicion to a genetic diagnosis is frequently prolonged. Families often describe a multi-year "diagnostic odyssey" across successive specialists before obtaining a genetic referral and a result[5]. Crucially, while the analytic performance of genomic platforms is extensively reported, the psychosocial and cultural mechanisms that determine whether families actually reach and engage with genetic evaluation remain comparatively fragmented across the literature.

Access to health services can be understood as the fit between the characteristics of services and the abilities of the people who seek them, spanning affordability, availability and accommodation, acceptability, approachability and appropriateness[6]. Within this framing, barriers are conditions that hinder, delay or prevent access[7], whereas facilitators are environmental, social or personal conditions that improve functioning and reduce the restrictions associated with disability[8]. For people with ID specifically, these general access dynamics are compounded within the clinical encounter by attitudinal and communicative barriers, including a priori assumptions of cognitive incapacity that can exclude the individual from decisions about their own care — a pattern that can be read as a form of epistemic injustice[9].

Understanding how these barriers and facilitators operate is essential to ensuring equitable genetic services for the ID population. Prior systematic reviews have addressed adjacent questions — factors acting as barriers to referral to genetic services[10], the opinions and experiences of people with ID regarding genetic testing and genetic medicine[11], and barriers and facilitators to cervical screening among people with ID[12] — but, to our knowledge, none has integrated the structural, sociocultural and psychosocial factors specifically along the genetic-evaluation pathway in ID, across a multi-informant evidence base, and none has foregrounded a Latin-American equity perspective. Accordingly, the objective of this review is to synthesise qualitative, mixed-methods and quantitative empirical evidence on the psychosocial and cultural barriers and facilitators influencing access to and engagement with the genetic-evaluation pathway among individuals with ID, their families and caregivers. Structured with the SPIDER framework[13], the review question asks: what are the psychosocial and cultural barriers and facilitators influencing access to and engagement with the genetic-evaluation pathway among individuals with intellectual disability — including associated neurodevelopmental or syndromic aetiologies — and their families or caregivers, according to empirical evidence published between 2020 and 2026?

2. Methods

2.1 Protocol and registration

This review is reported in accordance with the PRISMA 2020 statement[14]; the search is reported following PRISMA-S and the qualitative synthesis following ENTREQ[15]. During conduct, the scope was amended from an initial Latin-American focus to a global scope owing to a scarcity of eligible regional studies; this deviation is declared here. The review was not registered in a prospective systematic-review registry.

2.2 Eligibility criteria and framework

Eligibility was defined a priori using the SPIDER framework[13]. *Sample:* individuals with a primary or associated diagnosis of ID (or developmental disability) and their family members and caregivers. *Phenomenon of Interest:* psychosocial and cultural barriers and facilitators influencing access to, referral for, or uptake of genetic evaluation. *Design and Research type:* primary empirical studies using qualitative, quantitative or mixed-methods designs. *Evaluation:* subjective experiences, attitudes, perceptions, stigma, beliefs and socioeconomic constraints. Peer-reviewed journal articles published between January 2020 and March 2026 were eligible.

Exclusion criteria were: single-case designs without formal qualitative analysis; secondary literature (narrative and systematic reviews, meta-analyses); study protocols; editorials and expert opinion; and conference abstracts. The restriction to English-language publications was adopted for feasibility and is acknowledged as a limitation and a likely source of regional underrepresentation. Grey literature was excluded as a pragmatic decision; we note that this may introduce publication bias, since theses and reports can contain rich qualitative material.

2.3 Information sources and search strategy

A systematic search was executed in PubMed and Scopus on 13 March 2026. Search strings combined controlled vocabulary (MeSH in PubMed) with free-text keywords, using Boolean operators and truncation to capture linguistic variants. The exact strings as executed, with field tags (TITLE-ABS-KEY in Scopus) and the number of records retrieved per database, are reported in Supplementary Material so that the search is reproducible.

2.4 Study selection

Retrieved records were compiled in a shared repository, and 111 duplicate records identified across the two databases (by DOI) were removed. Titles and abstracts were screened against the eligibility criteria: the screening workload was distributed between the reviewers, and all excluded and uncertain records were cross-checked by the alternate reviewer, with disagreements resolved through discussion. Full texts of potentially eligible records were then assessed. The selection process and the number of records at each stage are summarised in the PRISMA 2020

flow diagram (Figure 1).

2.5 Data extraction

A standardised data-extraction matrix was developed and piloted. Extracted variables covered bibliographic metadata (authors, year, country of origin), methodological characteristics (design and data-collection instruments), sample details (size and participant roles) and thematic content (barriers, facilitators and principal conclusions). Study characteristics are presented in Table 1, and the full extraction matrix in Supplementary Table S1.

2.6 Quality appraisal

Methodological quality was appraised using the Mixed Methods Appraisal Tool version 2018 (MMAT)[16], applying the item set corresponding to each study's design category, independently in duplicate, with disagreements resolved by discussion. Consistent with MMAT guidance, studies were not excluded on the basis of appraisal; instead, quality informed the assessment of confidence and a sensitivity consideration.

2.7 Data synthesis

Findings were synthesised using thematic synthesis[17], conducted in three stages: line-by-line coding of the findings reported in the included studies; organisation of codes into descriptive themes; and development of analytical themes. The three analytical domains reported below — structural/systemic, sociocultural and psychosocial — were derived through this process rather than imposed a priori. Confidence in each review finding was assessed using the GRADE-CERQual approach[18] across its four components: methodological limitations, coherence, adequacy and relevance of the supporting data.

2.8 Ethics

Ethical approval was not required, as this review synthesised previously published studies and did not involve the collection of new data from human participants.

2.9 Use of AI-assisted tools

AI-assisted tools were used during the preparation of this manuscript; in accordance with the journal's policy this use is documented here. No AI tool met authorship criteria or is listed as an author, and the authors verified all references and factual claims against the primary sources and take full responsibility for the content.

3. Results

3.1 Included studies

The database search yielded 1,352 records (Scopus, 1,074; PubMed, 278); after removing 111 duplicates, 1,241 records were screened, and full-text assessment identified 20 studies meeting the eligibility criteria (one clinical case report and one study protocol were excluded per criteria; characteristics of the excluded studies are provided in Supplementary Material). Of these, **fourteen qualitative and mixed-methods studies** were included in the qualitative synthesis, while **six quantitative studies of access and uptake**[19–24] were retained as contextual evidence and are not synthesised here. The included studies were conducted across Australia, the United States, several European settings (including the United Kingdom and multinational European Reference Network structures), Israel, Japan and Brazil; publication dates ranged from 2020 to 2026. The study-selection process is summarised in the PRISMA 2020 flow diagram (Figure 1), and study characteristics in Table 1; confidence in each review finding is reported in Table 2 (GRADE-CERQual).

The included studies were methodologically diverse. Most used qualitative designs — semi-structured individual interviews, dyadic interviews and focus groups[25–28] — complemented by three mixed-methods studies[29–31]. Six quantitative survey and administrative-cohort studies of testing decisions and uptake[19–24] were retained as contextual evidence rather than synthesised. Participants reflected a multi-informant architecture: people with ID themselves, mothers, fathers and other family caregivers, clinicians and clinical geneticists, and secondary-school teachers. Full characteristics are reported in Table 1.

Findings were organised into two overarching categories — barriers and facilitators — each structured across three domains: structural/systemic, sociocultural and psychosocial. Many studies contributed to more than one domain; the number of studies supporting each finding, and the corresponding confidence in it, are reported in Table 2 (GRADE-CERQual).

3.2 Barriers

3.2.1 Structural and systemic barriers

Structural and systemic factors were the most frequently described domain in the qualitative corpus, and are further corroborated by contextual quantitative evidence of unequal test uptake.

Cost and insurance authorisation. Studies described the cost of testing and third-party authorisation as central obstacles. Simon et al.[5] documented administrative delays in which insurance pre-authorisation postponed testing — in some accounts for over three years — alongside clinical waitlists and limited initial provider validation (review finding B1; moderate confidence, Table 2). This qualitative account is corroborated by contextual quantitative evidence: two administrative-cohort studies reported lower rates of microarray or exome testing for children with ID alone than for children with co-occurring autism within a public insurance system[21] and variation in chromosomal-microarray use by socioeconomic and ethnic group[22], and a national

clinician survey linked the decision to test to families' anticipated out-of-pocket burden[20].

Workforce readiness and specialist access. In the qualitative corpus, provider unreadiness surfaced in families' accounts of referral delay and limited provider validation[5]. As contextual quantitative evidence, two national surveys of clinicians found that decisions to order genomic testing were associated primarily with the presence of overt dysmorphic features rather than with standardised criteria[19,20].

Fragmented pathways and educational exclusion. After diagnosis, families described discontinuities in care: Copeland et al.[29] reported a disconnect between receiving a molecular result and accessing practical, localised follow-up, and Kleinendorst et al.[27] documented the absence of definitive prognostic answers for susceptibility copy-number variants of variable expression (review finding B2; moderate confidence). Within the education system, teachers reported willingness to build genomic literacy in neurodivergent students but a lack of training and adapted resources[32]; and young people with ID reported that professionals often disregarded their input and did not communicate in accessible ways[33].

3.2.2 Sociocultural barriers

Six studies described sociocultural barriers, operating through stigma, assumptions of incompetence and culturally shaped meanings of disability.

Stigma and the reconfiguration of family identity. Receiving a molecular diagnosis was associated with a reconstruction of family identity and, at times, with social stigma. Fitzgerald et al.[26] described how a rare genetic diagnosis reshaped parents' sense of lineage and belonging, and how over-technical or unvalidated disclosure during feedback sessions was associated with parental alienation. In families of children with co-occurring autism and ID, Klitzman et al.[28] reported internalised parental blame, perceived social stigma and behavioural over-attribution following a molecular diagnosis (review finding B5; moderate confidence).

Assumptions of cognitive incompetence and paternalism. Several studies described an a priori assumption of incapacity that marginalised the person with ID within the clinical encounter. Strnadová et al.[25] characterised genetic-healthcare communication as over-medicalised and as marginalising patient autonomy. From the direct perspective of young people with ID, Hansen et al.[33] reported that professionals frequently disregarded their input and did not use accessible, age-appropriate communication, despite participants' stated desire for autonomy and genomic literacy. In secondary education, Jackaman et al.[32] linked teachers' constrained engagement with these topics to a lack of training and to low institutional expectations. This exclusion of people with ID from decisions about their own care was the most consistently supported barrier across the corpus (review finding B4; high confidence, Table 2).

Culturally shaped meanings of disability. One Brazilian study[34] described how religious and spiritual frameworks shaped caregivers' engagement: the same framing that provided emotional support (see 3.3.2) could, in some accounts, sustain a resignation that deferred proactive clinical follow-up. As this theme rests on a single study, it is of very low confidence and is advanced as

a hypothesis rather than a generalisable finding (review finding B7).

3.2.3 Psychosocial barriers

Seven qualitative studies described psychosocial barriers centred on caregiver distress, guilt and prognostic uncertainty.

Distress, anxiety and grief. Across studies, caregivers described grief and sustained distress. Fitzgerald et al.[26] reported grief and a reorientation of parental expectations following diagnosis. Luermans et al.[35] described fathers' initial emotional isolation followed by a pronounced need for structured informational support and future planning. Kleinendorst et al.[27] linked chronic parental distress to the variable phenotypic expression of susceptibility copy-number variants and to the absence of definitive prognostic information, and Klein Haneveld et al.[36] noted parental uncertainty regarding long-term developmental outcomes. Caregiver distress was among the most strongly supported findings (review finding B6; high confidence).

Guilt and biological responsibility. Klitzman et al.[28] reported internalised parental blame following a genetic diagnosis. In the context of positive newborn screening, Corbo et al.[37] described acute maternal distress and existential confusion driven primarily by the clinical ambiguity of premutation status, which could persist even when the identified variant might never become clinically significant (review finding B3; moderate confidence).

3.3 Facilitators

Fewer facilitators than barriers were identified, and those identified were supported with lower confidence (Table 2). This imbalance is interpreted, in part, as a property of the searched literature rather than as an established feature of the field, and is revisited in the Limitations.

3.3.1 Structural and systemic facilitators

Diagnostic resolution and integrated care. Reaching a molecular diagnosis was associated with clinical direction and with informing parental reproductive planning, although a gap to practical follow-up persisted[29] (review finding F5; low confidence). As contextual quantitative evidence, a single-centre cohort reported that an integrated, collaborative psychiatric-genetics care model was associated with shorter time-to-evaluation and fewer missed appointments[23].

Co-designed accessible information and clinician tools. Co-produced, accessible information designs (Easy Read format) were described as a promising facilitator (review finding F1; low confidence). Strnadová et al.[25] argued for accessible, co-produced information as a corrective to over-medicalised communication, and Kotwal et al.[30] developed and evaluated an Easy Read pre-clinic booklet; notably, this evaluation was conducted with clinical geneticists and genetic counsellors rather than with patients, so no reduction in patient anxiety or improvement in patient comprehension is claimed (see Future directions). At the workforce level, Strnadová et al.[31] reported that deployment of the co-produced GeneEQUAL toolkit was associated with

higher clinician self-rated capability, motivation and communication confidence: 96% of surveyed clinicians reported that the toolkit would improve the quality of their practice and 95% that it was effective (review finding F2; very low confidence, single study). Teachers, when provided with structured resources, were described as willing facilitators of early genomic literacy[32].

3.3.2 Sociocultural facilitators

One Brazilian study[34] described religious and spiritual coping as a resilience resource: primary maternal caregivers drew on faith and spiritual practices to buffer distress and to sustain long-term engagement with outpatient genetic services (review finding F4; very low confidence). Because this is the same study that supports the "religious resignation" barrier (3.2.2), the resignation–resilience tension is presented as a tentative, single-study observation, with the risk of double-counting declared explicitly.

3.3.3 Psychosocial facilitators

Two qualitative studies described how clear information and supportive dialogue were associated with reduced caregiver distress (review finding F3; low confidence). Corbo et al.[37] reported that resolving clinical uncertainty through dialogue with a genetic counsellor was associated with reassurance following the acute distress of a positive screen, and Fitzgerald et al.[26] linked validated, sensitive disclosure to more adaptive parental adjustment. Luermans et al.[35] similarly emphasised structured informational support. These studies describe the experience of caregivers rather than measured patient anxiety.

4. Discussion

This review synthesised empirical evidence on the psychosocial and cultural factors shaping access to and engagement with the genetic-evaluation pathway in intellectual disability (ID). Its principal finding is that this pathway is shaped by structural, sociocultural and psychosocial factors alongside technical ones: the prolonged "diagnostic odyssey" reported by families is not reducible to laboratory constraints or platform yields, but is co-produced by the organisation of services, the meanings attached to disability, and the emotional labour of caregiving[5,26,34].

The barrier–facilitator asymmetry is, in part, an artefact of the evidence base. Documented barriers outnumbered facilitators across the corpus. Rather than treating this as a property of the field, we interpret it cautiously as a feature of the searched literature: the search combined more barrier-oriented than facilitator-oriented terms, was restricted to two databases and to English, and the published literature tends to document deficits more readily than enablers. We therefore reframe the asymmetry as a limitation and a research priority rather than as a substantive conclusion.

Positioning relative to existing reviews. Contrary to a claim of complete novelty, several relevant syntheses precede this work: a systematic review of factors acting as barriers to referral

to genetic services[10]; a systematic review of the opinions and experiences of people with ID regarding genetic testing and genetic medicine[11]; and a systematic review and meta-analysis of barriers and facilitators to cervical screening in people with ID[12]. Our contribution is therefore delimited rather than absolute: we integrate structural, sociocultural and psychosocial factors specifically along the genetic-evaluation pathway in ID, across a multi-informant evidence base, and we foreground a Latin-American equity perspective that prior reviews did not address.

Exclusion from decision-making as epistemic injustice. A recurrent theme was the marginalisation of people with ID from decisions about their own genetic care: disregarded input, inaccessible communication, and a priori assumptions of incapacity[25,32,33]. This pattern can be read through the lens of testimonial injustice[9], in which a knower is granted diminished credibility on the basis of identity-based prejudice. Naming this mechanism reframes accessible, co-designed communication not as a courtesy but as a corrective to an epistemic harm — and locates the problem in professional and institutional practice rather than in the cognition of the person with ID.

A health-access framework for the findings. The results map coherently onto the five dimensions of patient-centred access described by Levesque et al.[6]: affordability (out-of-pocket cost and insurance authorisation[5,20]), availability and accommodation (specialist workforce and referral pathways[19,20,29]), acceptability (paternalism, stigma and assumptions of incapacity[25,28,33]), approachability (accessible information and outreach[30,31]) and appropriateness (continuity and post-diagnostic follow-up[27,29]).

A tentative cultural tension worth further study. A single Brazilian study[34] suggested that religious and spiritual frameworks can operate simultaneously as a resilience resource and as a source of resignation that defers proactive care. This duality is, to our knowledge, underexamined in the genetic-evaluation context and is analytically valuable; however, it rests on one study in one setting and requires replication before it can be generalised, particularly across Latin American contexts where extended family interdependence and religiosity are salient.

Actionable levers, advanced with graded confidence. Co-designed accessible information[25,30], clinician-capability toolkits (GeneEQUAL)[31], integrated care models[23] and educator professional development[32] emerged as promising levers. Their evidential base is uneven — several are single studies, and some were evaluated only with professionals rather than with patients — so we advance them as hypotheses for evaluation rather than as established interventions, with confidence rated accordingly (GRADE-CERQual).

The socioeconomic gradient. The administrative-cohort disparities in uptake[21,22] are consistent with genetic-evaluation access operating partly along socioeconomic and ethnic lines. We present this as an argued interpretation rather than as proof, and note that it derives from two specific health-system contexts (US Medicaid; the Israeli public system), which constrains generalisation.

5. Limitations

Several limitations qualify these findings. First, the search was restricted to two databases and to English-language publications; this is both a source of bias and the most plausible explanation for the barrier–facilitator asymmetry, and it partly manufactures the very Latin-American scarcity the review then reports, since excluding Spanish- and Portuguese-language literature under-captures regional evidence. Second, the corpus was methodologically heterogeneous: qualitative and mixed-methods studies formed the synthesis, whereas quantitative studies of access and uptake were treated as contextual evidence rather than synthesised. Third, several themes rest on single studies, limiting transferability. Fourth, several facilitator findings derive from one research programme (GeneEQUAL, UNSW), raising the possibility of participant overlap and warranting cautious interpretation. Fifth, the review was not prospectively registered, and the intermediate screening counts were reconstructed from the search logs and the per-database records. Finally, consistent with the GRADE-CERQual assessment, confidence in several findings — particularly single-study findings — is low to very low, and the conclusions are qualified accordingly.

6. Future directions

Priorities for future work follow directly from these limitations. First, primary empirical studies in Latin America — incorporating Spanish- and Portuguese-language literature and databases (e.g., LILACS, SciELO) — are needed to test whether the barriers described in high-income settings generalise, and to examine how macrostructural constraints intersect with regional sociocultural factors. Second, research should isolate ID from broad neurodevelopmental-disorder samples to chart its specific trajectories. Third, co-designed communication tools (Easy Read materials, clinician toolkits) should be evaluated with patient-level outcomes rather than only with professional endorsement. Fourth, the religious-coping duality warrants cross-cultural replication. Fifth, facilitator-focused and co-produced research is needed to correct the documentary imbalance identified here.

7. Conclusion

The pathway to genetic evaluation in intellectual disability is shaped by human, social and structural factors alongside technical ones. The current evidence is dominated by barriers — spanning cost and authorisation, workforce readiness, fragmented pathways, stigma, assumptions of incapacity, and caregiver distress — although whether facilitators are genuinely scarcer or merely under-studied remains an open question that this review cannot resolve. Co-designed communication, integrated care pathways and inclusive education emerge as actionable, if still tentatively evidenced, levers, and the systematic exclusion of people with ID from decisions about their own genetic care invites an epistemic-justice response rooted in professional and institutional change. Closing the Latin-American evidence gap is a priority that the present

review can name but not yet fill.

Data Availability Statement

All data supporting the findings are included in the article and its Supplementary Material (Supplementary Table S1).

Code availability. Not applicable; no custom code was used in the synthesis.

References

1. American Psychiatric Association. Diagnostic and statistical manual of mental disorders. 5th ed., text rev. American Psychiatric Association; 2022.
2. World Health Organization. International statistical classification of diseases and related health problems. 11th ed. (ICD-11). World Health Organization; 2019.
3. Schalock RL, Luckasson R, Tassé MJ, et al. Intellectual disability: definition, diagnosis, classification, and systems of supports. 12th ed. American Association on Intellectual and Developmental Disabilities; 2021.
4. Manickam K, McClain MR, Demmer LA, et al. Exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability: an evidence-based clinical guideline of the American College of Medical Genetics and Genomics. Genetics in Medicine. 2021;23(11):2029–2037. https://doi.org/10.1038/s41436-021-01242-6
5. Simon J, Hyde C, Saravanapandian V, Wilson R, Distefano C, Besterman A, et al. The diagnostic journey of genetically defined neurodevelopmental disorders. Journal of Neurodevelopmental Disorders. 2022;14(1):27. https://doi.org/10.1186/s11689-022-09439-9
6. Levesque J-F, Harris MF, Russell G. Patient-centred access to health care: conceptualising access at the interface of health systems and populations. International Journal for Equity in Health. 2013;12:18. https://doi.org/10.1186/1475-9276-12-18
7. Świgoń M. Information limits: definition, typology and types. Library Management. 2011;32(6/7):475–484. https://doi.org/10.1108/01435121111158619
8. World Health Organization. International classification of functioning, disability and health (ICF). World Health Organization; 2001.
9. Fricker M. Epistemic injustice: power and the ethics of knowing. Oxford University Press; 2007. https://doi.org/10.1093/acprof:oso/9780198237907.001.0001
10. Delikurt T, Williamson GR, Anastasiadou V, Skirton H. A systematic review of factors that act as barriers to patient referral to genetic services. European Journal of Human Genetics. 2015;23(6):739–745. https://doi.org/10.1038/ejhg.2014.180
11. Strnadová I, Loblinzk J, Scully JL, et al. The opinions and experiences of people with intellectual disability regarding genetic testing and genetic medicine: a systematic review. Genetics in Medicine. 2022;24(3):535–548. https://doi.org/10.1016/j.gim.2021.11.013
12. Power R, David M, Strnadová I, et al. Cervical screening participation and access facilitators and barriers for people with intellectual disability: a systematic review and meta-

analysis. Frontiers in Psychiatry. 2024;15:1379497. https://doi.org/10.3389/fpsyt.2024.1379497
13. Cooke A, Smith D, Booth A. Beyond PICO: the SPIDER tool for qualitative evidence synthesis. Qualitative Health Research. 2012;22(10):1435–1443. https://doi.org/10.1177/1049732312452938
14. Page MJ, McKenzie JE, Bossuyt PM, et al. The PRISMA 2020 statement: an updated guideline for reporting systematic reviews. BMJ. 2021;372:n71. https://doi.org/10.1136/bmj.n71
15. Tong A, Flemming K, McInnes E, Oliver S, Craig J. Enhancing transparency in reporting the synthesis of qualitative research: ENTREQ. BMC Medical Research Methodology. 2012;12:181. https://doi.org/10.1186/1471-2288-12-181
16. Hong QN, Fàbregues S, Bartlett G, et al. The Mixed Methods Appraisal Tool (MMAT) version 2018 for information professionals and researchers. Education for Information. 2018;34(4):285–291. https://doi.org/10.3233/EFI-180221
17. Thomas J, Harden A. Methods for the thematic synthesis of qualitative research in systematic reviews. BMC Medical Research Methodology. 2008;8:45. https://doi.org/10.1186/1471-2288-8-45
18. Lewin S, Glenton C, Munthe-Kaas H, et al. Using qualitative evidence in decision making for health and social interventions (GRADE-CERQual). PLoS Medicine. 2015;12(10):e1001895. https://doi.org/10.1371/journal.pmed.1001895
19. Okazaki T, Aoki C, Shinzato S, Adachi K, Nanba E. Awareness and knowledge of pediatricians regarding genetic testing for Fragile X syndrome in Japan: a national survey. Brain & Development. 2025;47(4):104367. https://doi.org/10.1016/j.braindev.2025.104367
20. Okazaki T, Aoki C, Adachi K, Yorifuji T, Hirasawa A, Nanba E. Japanese pediatric neurologist's decision regarding genetic testing for patients with developmental delay/intellectual disability: a nationwide survey. Brain & Development. 2025;47(3):104361. https://doi.org/10.1016/j.brain
21. Brown TR, Lee W-L, Ventimiglia J, Thurm A, Levy T, Yuan V, et al. Medicaid claims from 2008 to 2016 indicate low rates of genetic testing among children with intellectual disability and autism spectrum disorder. Genetics in Medicine. 2025;27(8):101451. https://doi.org/10.1016/j.gim.2025.101451
22. May D, Barshir R, Shahar M, Rose AJ, et al. Genetic testing of neurodevelopmental disorders in Israel. JAMA Network Open. 2025;8(8):e2527464. https://doi.org/10.1001/jamanetworkopen.2025
23. Shillington A, Lamy M, Dominick KC, Sorter M, Erickson CA, Hopkin R. A collaborative psychiatric-genetics inpatient care delivery model improves access to clinical genetic evaluation, testing, and diagnosis for patients with neurodevelopmental disorders. Frontiers in Genetics. 2022;13:901458. https://doi.org/10.3389/fgene.2022.901458
24. Pode-Shakked B, Barel O, Singer A, Regev M, Poran H, Eliyahu A, et al. A single center experience with publicly funded clinical exome sequencing for neurodevelopmental disorders or multiple congenital anomalies. Scientific Reports. 2021;11(1):19099. https://doi.org/10.1038/s41598-021-98646-w
25. Strnadová I, Loblinzk J, Scully JL, et al. "I am not a number!" Opinions and preferences of people with intellectual disability about genetic healthcare. European Journal of Human Genetics. 2023;31(9):1057–1065. https://doi.org/10.1038/s41431-023-01282-3
26. Fitzgerald J, Wilson C, Kelly C, Gallagher L. 'More than a box of puzzles': understanding the parental experience of having a child with a rare genetic condition. European Journal

of Medical Genetics. 2021;64(4):104164. https://doi.org/10.1016/j.ejmg.2021.104164

27. Kleinendorst L, van den Heuvel LM, Henneman L, van Haelst MM. Who ever heard of 16p11.2 deletion syndrome? Parents' perspectives on a susceptibility copy number variation syndrome. European Journal of Human Genetics. 2020;28(9):1196–1204. https://doi.org/10.1038/s41020-0644-6
28. Klitzman R, Bezborodko E, Chung WK, Appelbaum PS. Impact of receiving genetic diagnoses on parents' perceptions of their children with autism and intellectual disability. Journal of Autism and Developmental Disorders. 2025;55(1):284–296. https://doi.org/10.1007/s10803-023-06195-0
29. Copeland H, Kivuva E, Firth HV, Wright CF. Systematic assessment of outcomes following a genetic diagnosis identified through a large-scale sequencing study. Genetics in Medicine. 2021;23(6):1058–1064. https://doi.org/10.1038/s41436-021-01110-3
30. Kotwal H, Fleming J, Barlow-Stewart K, Boyle J, Silberbauer L, Leffler M, et al. Pre-genetics clinic resource evaluation for adults with intellectual disability: the pre-genetics clinic aid. Journal of Genetic Counseling. 2020;29(4):668–677. https://doi.org/10.1002/jgc4.1259
31. Strnadová I, Dunn M, Molnar C, et al. "All doctors should be trained in that": the co-production and mixed-methods evaluation of an educational toolkit to enable safe, high-quality genetic health care for people with intellectual disability (GeneEQUAL). Genetics in Medicine. 2025;27(4):101371. https://doi.org/10.1016/j.gim.2025.101371
32. Jackaman K-M, Strnadová I, Willow SA, et al. Developing genetic literacy in high school students with intellectual disability: teachers' experiences and perspectives. European Journal of Human Genetics. 2025;33(11):1530–1538. https://doi.org/10.1038/s41431-025-01865-2
33. Hansen J, Strnadová I, Danker J, Jackaman K-M, et al. "It was up to me to be curious": perceptions and experiences of students with intellectual disability on genetics and health education. European Journal of Human Genetics. 2026. Advance online publication. https://doi.org/10.1038/s41431-026-02041-w
34. de Araújo Rodrigues S, Zapparoli Nunes de Carvalho E, Campos CA, Barcellos Fontanella BJ, Gusmão Melo D. Exploring spiritual/religious coping strategies among mothers of children with severe or profound intellectual disability during genetic counseling in Brazil. Journal of Genetic Counseling. 2024;33(4):745–756. https://doi.org/10.1002/jgc4.1783
35. Luermans J, Fleming J, O'Shea R, Barlow-Stewart K, Palmer EE, Leffler M. "We are not a typical family anymore": exploring the experiences and support needs of fathers of children with a genetic condition. American Journal of Medical Genetics Part A. 2024;194(4):e63470. https://doi.org/10.1002/ajmg.a.63470
36. Klein Haneveld MJ, de Mortier CA, Hugon A, Cornel MC, et al. 'We are the engine': a focus group study on clinical practice guideline development with people with rare genetic neurodevelopmental disorders. Orphanet Journal of Rare Diseases. 2025;20(1):169. https://doi.org/10.1186/s13023-025-03673-9
37. Corbo A, Tzeng JP, Scott S, Cheves E, Cope H, Peay H. Parent perspectives following newborn screening resulting in diagnoses of fragile X syndrome or fragile X premutation. Research in Developmental Disabilities. 2024;148:104719. https://doi.org/10.1016/j.ridd.2024.104719

Acknowledgments

The authors thank the research support teams at Universidad CES.

Author Contributions

Conceptualization: S.M.H., S.G.V., P.V.-P.; Methodology: S.M.H., S.G.V., P.V.-P.; Investigation and study screening: S.M.H., S.G.V.; Formal analysis (thematic synthesis): S.M.H., S.G.V.; Data curation (extraction): S.M.H., S.G.V.; Writing – original draft: S.M.H., S.G.V.; Writing – review & editing: S.M.H., S.G.V., P.V.-P.; Supervision: P.V.-P. All authors read and approved the final manuscript, and all participated in the MMAT quality appraisal in duplicate and in resolving disagreements. *(S.M.H. = Sara Millán Huertas; S.G.V. = Sofía Gómez Vergara; P.V.-P. = Pedro Velez-Pardo.)*

Ethical Approval

Not required; the study synthesised previously published research and did not involve new data collection from human participants.

Funding

The authors received no specific funding for this work.

Competing Interests

The authors declare no competing interests.

Figures

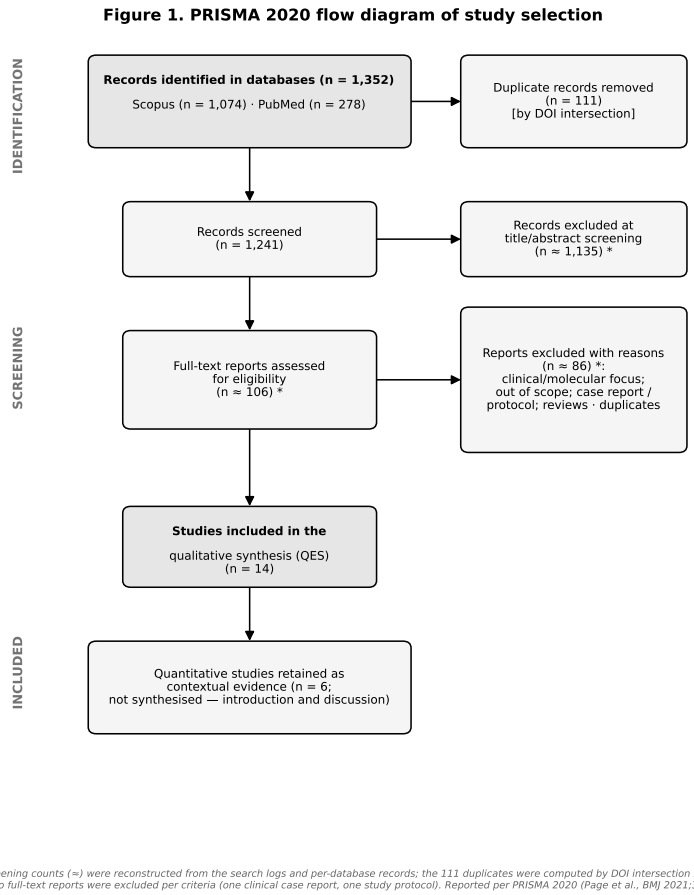


Figure 1. PRISMA 2020 flow diagram of study selection. Intermediate screening counts (≈) were reconstructed from the search logs and per-database records; the 111 duplicates were computed by DOI intersection between the raw exports.

Figure 2. Thematic map of barriers and facilitators to the genetic-evaluation pathway in intellectual disability

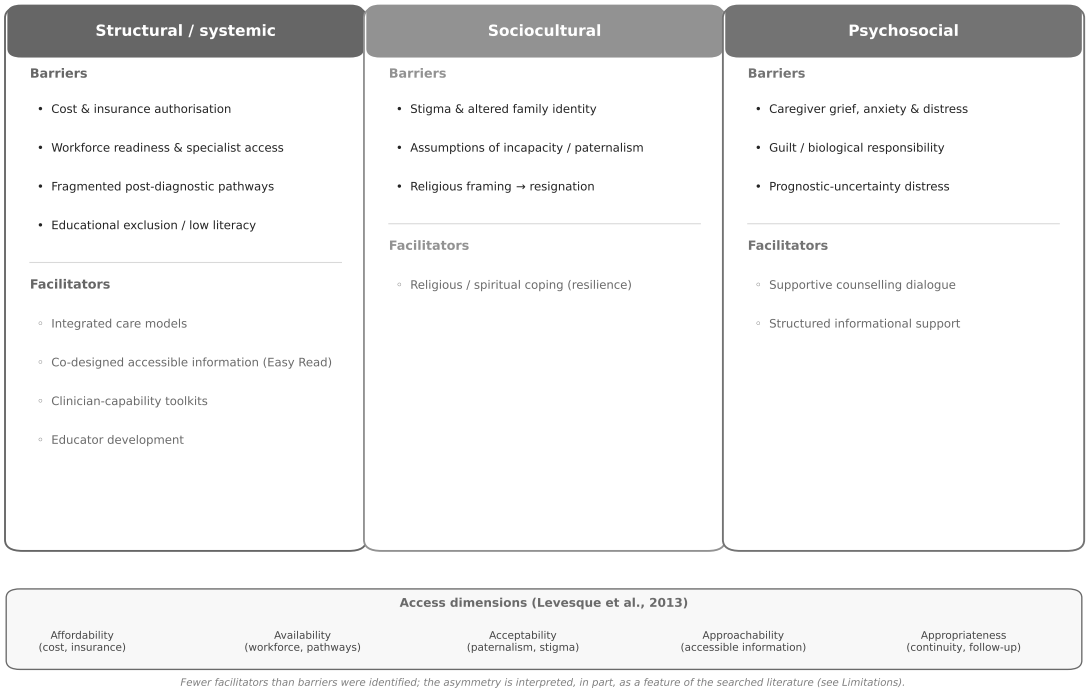


Figure 2. Thematic map of barriers and facilitators across the structural/systemic, sociocultural and psychosocial domains, mapped to the access dimensions of Levesque et al. (2013).

Figure 3. Evidence and confidence map of review findings (GRADE-CERQual)

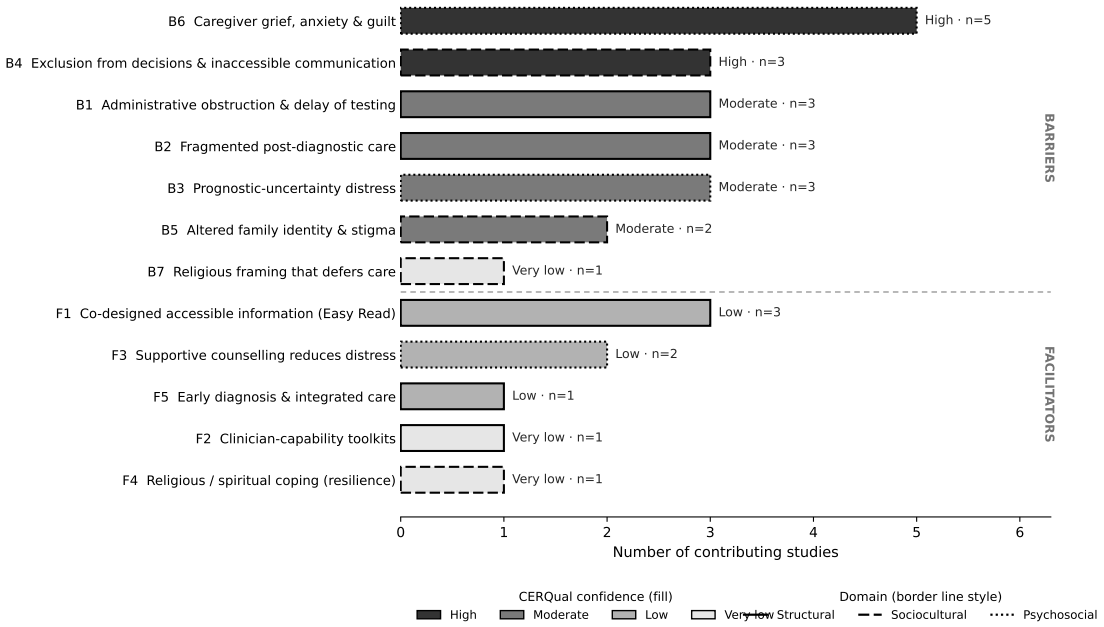


Figure 3. Evidence and confidence map of the review findings, showing the number of contributing studies (bar length), the GRADE-CERQual confidence (fill) and the domain (border line style).

Tables

Table 1: Characteristics of the included studies (n = 20: 14 synthesised, 6 contextual).

Study	Country	Design & data collection	Sample (N; participants)	Role	Domains (B barriers; F facilitators)
Klein Haneveld et al. (2025)	Europe	Qualitative — focus groups (inductive thematic analysis)	N=21 patient advocates (ERN ITHACA), mostly parents	Synthesis	B: Structural, Psychosocial; F: —
Strnadová et al. (2023)	Europe/Australia	Inclusive qualitative — multimodal interviews + validation groups	N=61: 28 adults with ID, 8 supporters, 25 stakeholders	Synthesis	B: Structural; F: Structural
Luermans et al. (2024)	Australia	Qualitative — semi-structured telephone interviews	N=11 fathers of children with fragile X	Synthesis	B: Structural, Psychosocial; F: Structural, Psychosocial
Copeland et al. (2021)	England	Mixed methods — DECIPHER audit + interviews	N=118: 112 probands (audit), 6 parents (interviews)	Synthesis	B: Structural, Psychosocial; F: Structural
Kleinendorst et al. (2020)	Europe	Qualitative — 3 focus groups + questionnaire	N=23 relatives of 16 families	Synthesis	B: Structural, Psychosocial; F: Structural
Fitzgerald et al. (2021)	Europe	Qualitative — individual/dyadic interviews (constructivist)	N=30 parents (24 mothers, 6 fathers)	Synthesis	B: Sociocultural, Psychosocial; F: Structural, Psychosocial
Simon et al. (2022)	USA	Descriptive qualitative — 'diagnostic journey' interviews	N=37 families/caregivers of people with genetically defined NDD	Synthesis	B: Structural; F: —
Jackaman et al. (2025)	Australia	Qualitative — 11 interviews + 1 focus group	N=15 secondary-school teachers	Synthesis	B: Sociocultural, Structural; F: Structural
Klitzman et al. (2025)	USA	Qualitative — semi-structured interviews	N=28 parents (15 mothers, 13 fathers) of children with autism + ID	Synthesis	B: Sociocultural, Psychosocial; F: —

Study	Country	Design & data collection	Sample (N; participants)	Role	Domains (B barriers; F facilitators)
Hansen et al. (2026)	Australia	Inclusive qualitative — Easy Read interviews	N=14 students/young people with ID (12–28 y)	Synthesis	B: Sociocultural, Structural; F: —
Kotwal et al. (2020)	Australia	Mixed-methods feasibility pilot (co-design + REDCap)	N=69: 3 co-design (2 with ID), 66 professionals	Synthesis	B: —; F: Structural
Strnadová et al. (2025)	Australia	Mixed-methods evaluation — COM-B, interviews, focus group	N=115: 78 clinicians, 21 interviews, 15 with ID + 1 supporter	Synthesis	B: —; F: Structural
Araújo Rodrigues et al. (2024)	Brazil	Clinical-descriptive qualitative — home interviews	N=15 mothers/caregivers (>6 months adherence)	Synthesis	B: Sociocultural; F: Sociocultural
Corbo et al. (2024)	USA	Qualitative — semi-structured interviews	N=24 mothers after newborn screening (6 fragile X, 18 premutation)	Synthesis	B: Psychosocial; F: Psychosocial
Okazaki et al. (2025a)	Japan	National cross-sectional survey (online questionnaire)	N=326: 78 clinical geneticists, 248 non-geneticist paediatricians	Context	B: Structural; F: —
Okazaki et al. (2025b)	Japan	National cross-sectional survey	N=266 neuropaediatricians (41 also geneticists)	Context	B: Structural; F: —
Brown et al. (2025)	USA	Retrospective cohort — Medicaid claims 2008–2016	N=241,060 (ID, ASD, ASD+ID, controls)	Context	B: Sociocultural, Psychosocial; F: Structural
May et al. (2025)	Israel	Retrospective population cohort (EHR)	N=25,403 patients with NDD (public network)	Context	B: Structural; F: —
Shillington et al. (2022)	USA	Retrospective cohort — EHR review	N=135: 34 pre, 101 post collaborative model	Context	B: Structural; F: —
Pode-Shakked et al. (2021)	Israel	Retrospective single-centre cohort — clinical exome	N=280 undiagnosed probands (funded pathway)	Context	B: Structural; F: —

Study	Country	Design & data collection	Sample (N; participants)	Role	Domains (B barriers; F facilitators)

Table 2: Summary of qualitative findings and GRADE-CERQual confidence assessment.

Finding	Review finding	Contributing studies (n)	Methodological limitations	Coherence	Adequacy	Relevance	CERQual confidence
B1	The genetic-evaluation pathway is experienced as administratively obstructed and prolonged (waits, insurance pre-authorisation)	Simon, Copeland, Klein Haneveld (3)	Minor	Coherent	Moderate	High	Moderate
B2	Disconnect between the molecular result and practical care/follow-up	Copeland, Kleinendorst, Klein Haneveld (3)	Minor to moderate	Coherent	Moderate	High	Moderate
B3	Prognostic ambiguity (variants of variable expression) sustains distress	Kleinendorst, Corbo, Klein Haneveld (3)	Minor	Coherent	Moderate	High	Moderate
B4	People with ID are excluded from decisions about their own care and addressed in inaccessible ways (paternalism, presumption of incapacity)	Strnadová 2023, Hansen, Jackaman (3)	Minor	Coherent	Rich (direct voice)	High	High
B5	A molecular diagnosis reconfigures family identity and can bring stigma	Fitzgerald, Klitzman (2)	Minor	Coherent	Moderate	High	Moderate
B6	Grief, anxiety and caregiver guilt following diagnosis	Fitzgerald, Luermans, Klitzman, Corbo, Kleinendorst (5)	Minor	Coherent	Rich	High	High
B7	Religious/cultural framing may sustain a resignation that defers proactive care	Araújo Rodrigues (1)	Minor	n/a (single study)	Limited	High (regional)	Very low

Finding	Review finding	Contributing studies (n)	Methodological limitations	Coherence	Adequacy	Relevance	CERQual confidence
F1	Co-designed accessible information (Easy Read) is valued as improving communication	Strnadová 2023, Kotwal, Strnadová 2025 (3)	Moderate (evaluated with professionals, not patients)	Coherent	Moderate	Partial (patient outcome absent)	Low
F2	Clinician-capability toolkits improve self-perceived inclusive practice	Strnadová 2025 (1)	Moderate (self-report)	n/a (single study)	Limited	Partial	Very low
F3	Counselling dialogue and clear information reduce caregiver distress	Corbo, Fitzgerald (2)	Minor	Coherent	Moderate	High	Low
F4	Religious/spiritual coping functions as a resilience resource	Araújo Rodrigues (1)	Minor	n/a (single study)	Limited	High (regional)	Very low
F5	Early diagnostic resolution and integrated care models facilitate navigation	Copeland (+ context: Shillington) (1-2)	Moderate	Coherent	Limited	High	Low

Supplementary Files

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- SupplementaryMMAT.xlsx