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Healthcare professionals' experiences on the communication of genetic information from parents to their young children: a scoping review protocol

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ADMINISTRATIVE INFORMATION

Support - FCT.

Review Stage at time of this submission - Data analysis.

Conflicts of interest - None declared.

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Amendments - This protocol was registered with the International Platform of Registered Systematic Review and Meta-Analysis Protocols (INPLASY) on 03 November 2023 and was last updated on 13 November 2023.

INTRODUCTION

Review question / Objective This scoping review aims to map the evidence on healthcare professionals' (HCPs) experiences with the communication of genetic information from parents to their young children (≤ 24 years).
Review questions: (1) What are HCP's experiences regarding the communication of genetic information from parents to their young children (≤24 years)?; and (2) What challenges, barriers and facilitators are described by HCP when addressing the communication of genetic information from parents to their children (≤24 years)?

Background Inherited genetic conditions (IGC) result from pathogenic variants in genes that can be transmitted to offspring (1). Genetic testing enables the detection of these pathogenic variants in blood relatives of individuals affected by the IGC or in carriers of these genetic variants. Carriers of pathogenic gene variants face an augmented risk of developing the IGC and/or passing them onto their offspring.
Communication plays an important role in how families manage and adjust to living with IGC (2). Parents often discuss the existing impacts of IGC on their children or inform them about the genetic risk (3). The literature indicates that parents often struggle with balancing the desire to protect their children from potentially distressing information and fostering open communication to ensure a comprehensive understanding of genetic risk and IGC while providing necessary emotional support (4,5). This communication may also be complicated by parents' ongoing adaptation to their own genetic status or IGC, feelings of guilt regarding potential genetic transmission, and concerns about their children's capacity to grasp complex genetic information (6). These discussions commonly present challenges for parents, prompting them to seek guidance and support from healthcare professionals (HCPs) on optimal

timing, approaches, and language for such conversations (7).

In genetic counseling, healthcare professionals (HCPs) assist individuals in comprehending and adjusting to the medical, psychological, and familial implications of IGCs. Guidelines recommend that HCPs actively encourage and support parents to disclose genetic risk information to their children, or engage in open discussions about IGCs as early as reasonably possible (4). HCPs serve as gatekeepers of genetic knowledge, guiding parents in effectively approaching these conversations (3). However, there is a lack of clarity regarding the role of HCPs in supporting parents in communicating genetic information to their young children, and there is limited research describing the experiences of HCPs in facilitating this communication.

This scoping review aims to address this research gap by mapping the evidence on the experiences of HCPs regarding the communication of genetic information between parents and young children (≤ 24 years).

1. Wakap, S., Lambert, D.M., Olry, A., Rodwell, C., Gueydan, C., Lanneau, V., et al. (2020). Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. European Journal of Human Genetics, 28, 165–173. https://doi.org/10.1038/s41431-019-0508-0

2. Rowland, E., & Metcalfe, A. (2013). Communicating inherited genetic risk between parent and child: a meta-thematic synthesis. International Journal of Nursing Studies, 50, 870–880. https://doi.org/10.1016/j.ijnurstu.2012.09.002

3. Etchegary, H., & Fowler, K. (2008). 'They had the right to know': genetic risk and perceptions of responsibility. Psychology and Health, 23, 707–727. https://doi.org/10.1080/14768320701235249

4. Forrest Keenan, K., Finnie, R. M., Simpson, W. G., McKee, L., Dean, J., & Miedzybrodzka, Z. (2019). Parents' views of genetic testing and treatment of familial hypercholesterolemia in children: A qualitative study. Journal of Community Genetics, 10, 129–141. https://doi.org/10.1007/s12687-018-0373-5

5. Bush, L.W., Bartoshesky, L.E., David, K.L., Wilfond, B., Williams, J.L., & Holm, I.A. (2018). Pediatric clinical genome sequencing and the engagement process: encouraging active conversation with the older child and adolescent: points to consider-a statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 20, 692–694. https://doi.org/10.1038/gim.2018.36

6. Middleton, J., Calam, R., & Ulph, F. (2018). Communication with children about sickle cell disease: a qualitative study of parent experience. British Journal of Health Psychology, 23, 685–700. https://doi.org/10.1111/bjhp.12311

7. Plumridge, G., Metcalfe, A., Coad, J., & Gill, P. (2011). Parents' communication with siblings of children affected by an inherited genetic condition. Journal of Genetic Counseling, 20, 374–383. https://doi.org/10.1007/s10897-011-9361-1

Rationale Genetic counseling guidelines advocate for HCPs to actively encourage parents to disclose genetic risk information to their children, or engage in open discussions about IGCs as early as reasonably possible. HCPs play a crucial role in this process, guiding and supporting parents navigate through this communication (3). Nevertheless, there remains uncertainty about the specific support HCPs should offer to parents in communicating genetic information to their young children. Moreover, there is limited literature on the experiences of HCPs in facilitating this communication.

This scoping review centers the perspective of HCP working with people at risk or with IGCs. A variety of IGCs will be considered, with different inheritance patterns, age at onset, morbidity, and life expectancy.

METHODS

Strategy of data synthesis Searched databases included Scopus, Web of Science, PubMed and PsycINFO. For PubMed and PsycINFO, an advanced search on title and abstracted was conducted. Web of Science and Scopus were searched for keywords.

Query strings in each database: ("health professional*" OR "healthcare professional*" OR "healthcare provider*" OR clinician* OR "healthcare practitioner*") AND ("parent-child*" OR "parent*" OR "offspring*" OR "minor*" OR "child*" OR "teen*" OR "adolescent*" OR "young adult*" OR "infant*" OR "relative*") AND ("genetic* disease*" OR "genetic* condition*" OR "inherit* disease*" OR "inherit* condition*" OR "genetic* information" OR "genetic* risk*" OR "hereditar*" OR "genetic counsel*") AND ("need*" OR "barrier*" OR "challenge*" OR "inform*" OR "transmit*" OR "facilitat*" OR "disseminat*" OR "communicat*" OR "shar*" OR "disclos*").

Eligibility criteria Inclusion criteria: Original peer-reviewed empirical studies published since 1997 in English, Portuguese, Spanish and French; empirical research studies reporting quantitative, qualitative and mixed-methods studies;

Population: HCP (e.g., physicians, psychologists, nurses, social workers);

Concept: experiences (practice, roles, challenges, and barriers and facilitators regarding the

communication of genetic information from parents to young children (≤24 years);
Context: all healthcare contexts addressing the communication of genetic information between parents and children;
Exclusion criteria: Studies published before 1997; studies not published in English, Portuguese, Spanish and French; studies that were not peer-reviewed; secondary research (e.g. literature reviews, systematic reviews, scoping reviews, opinion papers and books); studies for which the full-text was not available;
Population: Parents' and/or children's views, without providing the perspective of HCP;
Concept: Children's views on genetic testing uptake or parental views on prenatal testing or carrier testing of their young children; focus on interventions or reporting impacts or outcomes of parent-children communication; studies not focusing specifically on parent-children communication.

Source of evidence screening and selection Duplicate records were first removed using Zotero. Next, an initial screening of the records was carried out based on their title and abstract. A second screening involved reviewing the full text of the papers. Two independent reviewers conducted the screening, and any disagreement was resolved by involving a third reviewer.

Data management Data were managed using Zotero and Microsoft Office (Word). Zotero served to remove duplicates and screening. Microsoft Office (Word) was used to synthesize results. All sources and decisions were made visible using this software.

Reporting results / Analysis of the evidence Data will be analyzed in accordance with descriptive content analysis (since it is a scoping review) (1). Synthetized findings will report following PRISMA-ScR (2).

1. Pollock, D., Peters, M. D. J., Khalil, H., McInerney, P., Alexander, L., Tricco, A. C., Evans, C., De Moraes, É. B., Godfrey, C., Pieper, D., Saran, A., Stern, C., & Munn, Z. (2022). Recommendations for the extraction, analysis, and presentation of results in scoping reviews. JBI Evidence Synthesis, 21(3), 520–532. https://doi.org/10.11124/jbies-22-00123
2. Tricco, A. C., Lillie, E., Zarin, W., O'Brien, K. K., Colquhoun, H., Levac, D., Moher, D., Peters, M. D. J., Horsley, T., Weeks, L., Hempel, S., Akl, E. A., Chang, C., McGowan, J., Stewart, L., Hartling, L., Aldcroft, A., Wilson, M. G., Garritty, C., . . . Straus, S. E. (2018). PRISMA Extension for Scoping Reviews (PRISMA-SCR): Checklist and explanation. Annals of Internal Medicine, 169(7), 467–473. https://doi.org/10.7326/m18-0850

Presentation of the results This scoping reviews results will be presented according with the Preferred Reporting Items for Systematic Reviews and Meta-Analysis extension for scoping reviews (PRISMA-ScR) (1). Data will be presented also in tables and text.

1. Pollock, D., Peters, M. D. J., Khalil, H., McInerney, P., Alexander, L., Tricco, A. C., Evans, C., De Moraes, É. B., Godfrey, C., Pieper, D., Saran, A., Stern, C., & Munn, Z. (2022). Recommendations for the extraction, analysis, and presentation of results in scoping reviews. JBI Evidence Synthesis, 21(3), 520–532. https://doi.org/10.11124/jbies-22-00123

Language restriction Studies conducted in other languages than English, Portuguese, Spanish and French will not be included.

Country(ies) involved his scoping review was conducted in Portugal.

Keywords genetic counselling; genetic risk; healthcare professionals; hereditary disease; parent-children communication; young children.

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