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Communicating Genetic Information in Families with Inherited Late-Onset Neurodegenerative Diseases: A Scoping Review

Sofia Fontoura Dias[a,b], Maria Barbosa[c,d,e,f], Filipa Júlio[g,h,i,j], Angus Clarke[k], Alison Metcalfe[l], Jorge Sequeiros[c,d,m], Milena Paneque[c,d,m], Liliana Sousa[a,b], and Álvaro Mendes[c,d]

[a]Department of Education and Psychology, University of Aveiro; [b]CINTESIS@RISE–Centre for Health Technology and Services Research, University of Aveiro; [c]i3S – Instituto de Investigação e Inovação em Saúde, University of Porto; [d]CGPP – The Centre for Predictive and Preventive Genetics, IBMC – Institute for Molecular and Cell Biology, University of Porto; [e]FPCEUP – Faculty of Psychology and Educational Sciences, University of Porto; [f]CPUP – Centre for Psychology at the University of Porto; [g]European Huntington Association, Moerbeke-Waas; [h]Portuguese Huntington Association, Lisbon; [i]Faculty of Psychology and Educational Sciences, University of Coimbra; [j]CIBIT – Coimbra Institute for Biomedical Imaging and Translational Research, University of Coimbra; [k]Institute of Medical Genetics, School of Medicine, Cardiff University; [l]LOHA Health Ltd, Ludgate Hill; [m]ICBAS School of Medicine and Biomedical Sciences, University of Porto

ABSTRACT

Communicating genetic information within families living with inherited genetic conditions (IGCs), especially incurable and progressive late-onset neurodegenerative diseases (LONDs), presents significant challenges. To date, no literature review has specifically addressed this issue in families with LONDs. To fill this gap, a scoping review was conducted following PRISMA and JBI guidelines. Four databases (Scopus, Web of Science, PubMed, and PsycInfo) were searched, resulting in 31 articles for analysis. Six categories were identified: (i) from whom to whom: communication transmitters and receivers, (ii) what to tell: content and details of communication, (iii) how to tell: communication approach, (iv) when to tell: timing of communication, (v) barriers and reasons for not communicating, and (vi) facilitators and reasons for communicating. Sharing information about LONDs is a multi-step, deliberative process involving several transmitters and receivers. It typically begins with first-degree relatives, with women often assuming a pivotal role in the gathering and dissemination of information, and extends to the wider family. This process includes assessing the family member's subjective experiences and the potential impact of the information. Fear of stigmatization and discrimination are notable barriers, while feelings of responsibility and moral obligation toward family members are strong motivators for transmitters to communicate. The studies included in this review were all retrospective, primarily utilized a qualitative design, and predominantly focused on families with Huntington's disease. Prospective studies are needed to investigate disclosure decision-making in families with other LONDs and to explore how the potential availability of clinical trials for testing new drugs might influence family communication.

Introduction

When an inherited genetic condition (IGC) is diagnosed in an individual, family members may also have an interest in the information. Blood relatives may be at risk of developing the condition themselves and/or transmitting it to their offspring. Informed at-risk relatives can make considered decisions about undergoing genetic counseling and testing, act in advance to prevent future disease, and make more informed decisions about reproductive and other life-planning decisions. With genetic and genomic testing increasingly utilized in healthcare (McNeill, 2022) and direct-to-consumer testing (Horton et al., 2019), incorporating genetic information into routine family health communication is likely to become more common.

Informing relevant family members about an IGC is generally encouraged in genetic counseling. The responsibility for this communication typically falls to the first family member tested, the proband (Phillips et al., 2021). Family communication about an IGC involves multigenerational information management (Gaff & Bylund, 2010). This may include communicating about family health history (Campbell-Salome et al., 2019), disease characteristics and implications, such as the genetic risk for relatives and future offspring (Mendes et al., 2018; Rolland & Williams, 2005), as well as clinical trials (Andrew & Fox, 2023).

Although patients who receive genetic healthcare generally recognize the importance of communicating with their family members about the IGC, this communication is often challenging. The extent of this communication is influenced by various factors, including individual, interpersonal, and social factors, as well as disease characteristics (Ahsan et al., 2023; Gaff et al., 2007; Rowland & Metcalfe, 2013; Seymour et al., 2010; Srinivasan et al., 2020; Wilson et al., 2004; Wiseman et al., 2010). This process is also shaped by family dynamics and communication patterns, the degree of emotional closeness or distance, the perceived relevance of the information, the perceived recipient reactions, and the psychological burden of coping with their one's own results (Gomes

CONTACT Álvaro Mendes alvaro.mendes@ibmc.up.pt i3S – Instituto de Investigação e Inovação em Saúde, Universidade do Porto, Porto, Portugal
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et al., 2022; Mendes et al., 2018; Smit et al., 2021). Individuals often disclose genetic information out of a sense of responsibility or obligation toward others in the family, to obtain further information, to seek emotional support, or to enable family members to pursue in medical care and clarify their risk status (Gaff et al., 2007; Greenberg & Smith, 2015). This is a deliberative process that takes place within the context of family relationships, involving decisions about who should be informed, how the information will be conveyed, and anticipating the effects of (non)disclosure on one's own life and the lives of others (Ersig et al., 2011; Gaff & Bylund, 2010; Gaff et al., 2007).

In families living with late-onset neurodegenerative diseases (LONDs), communicating about the condition can be particularly challenging and complex, due to the progressive nature of the disease, its implications for future generations, and the emotional and psychological burdens associated with the diagnosis. LONDs, such as Huntington's disease (HD), the dominant spinocerebellar ataxias, and early-onset familial Alzheimer's disease, encompass a set of dominantly inherited, progressive, and incurable adult-onset conditions. These conditions often lead to severe impairment of movement, cognition, and behavior (Crook et al., 2022). Unlike IGCs such as hereditary cancer syndromes, which have effective prevention strategies and treatments capable of reducing morbidity and mortality, effective therapeutic options for LONDs are limited in preventing or significantly slowing disease progression. In some cases, the treatments may work only incompletely or work for a subset of patients (Crook et al., 2022). The lack of prevention and effective medical treatment is described as an additional barrier to family communication (Dheensa et al., 2016; Forrest et al., 2003; Wilson et al., 2004).

Previous literature reviews have described the process and outcomes of communicating genetic information within families living with various IGCs (Gaff et al., 2007; Rowland & Metcalfe, 2013; Wilson et al., 2004), and specifically in families with hereditary cancer syndromes (Seymour et al., 2010; Wiseman et al., 2010). They have also examined the efficacy of interventions aimed at improving the sharing of genetic test results within families (Ballard et al., 2023), investigated theoretically based approaches to foster family genetic risk communication (Zhao et al., 2022), and identified the barriers and facilitators to family communication following genetic testing for hereditary cancer syndromes (Ahsan et al., 2023; Seymour et al., 2010; Srinivasan et al., 2020). A recent integrative review developed a conceptual framework that elaborates the process of family information management in the context of IGCs, consisting of contextual influences, family information management behaviors, and family information management outcomes (Abad et al., 2024). Wiseman et al. (2010) identified Family Systems Theory as the most frequently used conceptual framework in studies on family communication about IGCs. The authors highlighted how family scripts, roles, and narratives significantly shape the sharing of genetic information within families. They particularly linked these communication dynamics to societal constructions of family and gender, emphasizing the expectations placed on women as primary caregivers and "kin keepers" (Wiseman et al., 2010). Family Systems Theory views families as interconnected and interdependent systems in which the behavior of each member influences and is influenced by others (McDaniel et al., 2006). It highlights that families function as emotional and relational units with unique communication patterns that are embedded in and shaped by wider contexts such as culture, along with intergenerational family dynamics, including belief systems, family roles, rules and boundaries, coping mechanisms, and life cycle challenges (McDaniel et al., 2006). According to the Family Systems Genetic Illness model (Rolland & Williams, 2005), the specific characteristics of IGCs (such as the likelihood of disease development, expected clinical severity, and timing of onset) give rise to distinct communication demands. These are likely to result in practical, emotional, and relational challenges for families that are unique to each condition (Rolland & Williams, 2005). A family systems approach has been employed to inform the development of tools to enhance understanding of patients' family dynamics (J. A. Peters et al., 2006), guiding interventions to promote family communication and social connectedness among families with IGCs, particularly through narrative (Stopford et al., 2020) and multifamily interventions (Guerra et al., 2023), and supporting the training of genetic counselors (Eisler et al., 2017).

Overall, the existing reviews primarily examined studies that focused on hereditary cancer syndromes. However, to our knowledge, no literature review has specifically addressed the communication of genetic information in families with LONDs. To address this gap, we conducted a scoping review to map existing research and identify gaps in the literature regarding the communication of genetic information in families affected by LONDs. Our primary research question was: "What is the available research evidence regarding the communication of genetic information in families with LONDs?;" the secondary research questions were: (1) "What are the key characteristics that guide the communication of genetic information in families with LONDs?" and (2) "What are the main barriers and facilitators in this communication?"

Materials and methods

This review was conducted following the Joanna Briggs Institute (JBI) guidance for scoping reviews (M. D. J. Peters et al., 2020) and is reported by the PRISMA extension for Scoping Reviews (PRISMA-ScR) (Tricco et al., 2018). Scoping reviews are a well-established method to identify key characteristics or factors of a given concept and research gaps (Munn et al., 2018; M. D. J. Peters et al., 2015). The protocol of this review was registered in the Open Science Framework (https://osf.io/cng36).

The Population, Concept, Context (PCC) approach was used to outline inclusion and exclusion criteria (Table 1):

Inclusion criteria also comprised primary peer-reviewed articles written in English, Portuguese, Spanish, or French published from 1997 onwards, since this was when direct mutation detection for HD became available and pre-symptomatic testing was widely adopted (Tibben, 2007). All types of research approaches were considered.

Table 1. Eligibility criteria according to the PCC approach.

Inclusion	Exclusion
Population Individuals living in families with a LOND, either at risk or affected, and their relatives.	Individuals living in families with IGC that are not a LOND, either at risk or affected and their relatives.
Concept Communication of genetic information (content, process of dissemination, and/or actions involved).	Only communication regarding genetic testing. Health care professionals' perspectives and experiences with facilitating family communication of genetic risk and decisions regarding genetic testing.
Context Studies in all contexts where family (nuclear or/and extended) communication of genetic risk information was addressed.	Studies addressing communication of genetic information in other contexts, e.g., social networks, healthcare professionals.

Search strategy

A search strategy was devised with the assistance of an academic librarian. An initial search on PubMed was conducted to explore MeSH terms and identify relevant English-language search terms aligned with the research questions for use in the search strategy. The same derived search terms were applied in all database searches, using the same filters and date ranges. Search terms included "genetic," "disclosure," "communication," "family," and "late-onset neurological diseases." The selection of the specific LONDs for the inclusion in the search was informed by the work of Crook et al. (2022), which is a systematic review of genetic counseling and testing practices for LONDs. Four online databases (Scopus, Web of Science, PubMed, and PsycInfo) were searched during July 2023. The search was supplemented by conducting manual citation searching of relevant papers. The full search strategy is available as Supplementary Material.

The articles were imported to Endnote 21 (Clarivate Analytics, PA, USA), and the duplicated records were first discarded using Endnote 21 and then imported to Rayyan (Qatar Computing Research Institute, Doha, Qatar). Each title and abstract was screened independently by the first and second authors. The second round of screening involved examining papers based on full texts by the same two independent reviewers. Discrepancies that emerged during the full-text screening were resolved via discussion with a third reviewer not involved in the initial decision (last author) to come to a consensus. All members of the review team are native Portuguese speakers, fluent in both English and Spanish. Additionally, the second author, who conducted the screening of all articles, is fluent in French.

Data extraction

A spreadsheet for extraction was created using Microsoft Excel to retrieve data from the included studies. One reviewer (first author) carried out the data extraction, while a second reviewer verified both its completeness and accuracy (second author). The data extracted (when available) concerned the study details (authors and year of publication), country of origin and language, the study aim(s), participants (sample size, disease status, sociodemographic and family characteristics), methods (study design and approach), IGC, and main findings about family communication of genetic information. The guidance sheet for data extraction is provided in Table S1 (supplementary material). The participants of the reviewed studies were categorized based on their disease status: (i) "affected" refers to individuals that are symptomatic carriers of the pathogenic gene variant; (ii) "pre-symptomatic carriers" are asymptomatic individuals identified as carriers of the pathogenic gene variant; (iii) "at-risk individuals (untested or with unknown test results/risk status)" includes offspring or other close relatives of affected individuals or pre-symptomatic carriers who have not undergone genetic testing or whose risk status is not specified in the study; (iv) "non-carriers" comprises individuals who have been tested and found not to carry the pathogenic gene variant; (v) "non-biological relatives" refers to family members not included in the biological genetic risk network, such as parents, aunts, uncles, grandparents on the unaffected side of the family, spouses, and in-laws.

Data analysis

The data were analyzed through descriptive statistical analysis for study characteristics and basic qualitative content analysis for textual data (Pollock et al., 2023). An inductive approach was utilized, focusing on identifying patterns and categories directly from the data, which informed the development of a framework comprising these categories. The content analysis involved familiarization with the data sources and an initial open coding phase. During this process, concepts or characteristics were identified and organized into overall categories (Pollock et al., 2023). After establishing a coding framework, which was refined and revised collaboratively by all members of the review team (first, second, and last authors) and three additional coauthors, the extracted data was organized according to this coding structure (M. D. J. Peters et al., 2020; Pollock et al., 2023). The main findings were subsequently extracted from the original text of all selected studies and organized within the established coding framework (Pollock et al., 2023). A summary of the synthesized findings in a narrative form was provided. In line with the established guidelines (M. D. J. Peters et al., 2020; Tricco et al., 2018), a quality rating of the studies and an assessment of the risk of bias were not conducted.

Results

The search yielded 5075 records as potential articles for inclusion. After removing duplicates, 2965 articles were screened based on their titles and abstracts. Three articles were manually added through citation searching. After excluding irrelevant articles, 34 publications were retrieved for full-text assessment of eligibility. In total, 31 articles met the eligibility criteria and were included in the review. The PRISMA flowchart detailing this process is presented in Figure 1. Detailed evidence regarding the included studies is presented in Table 2.

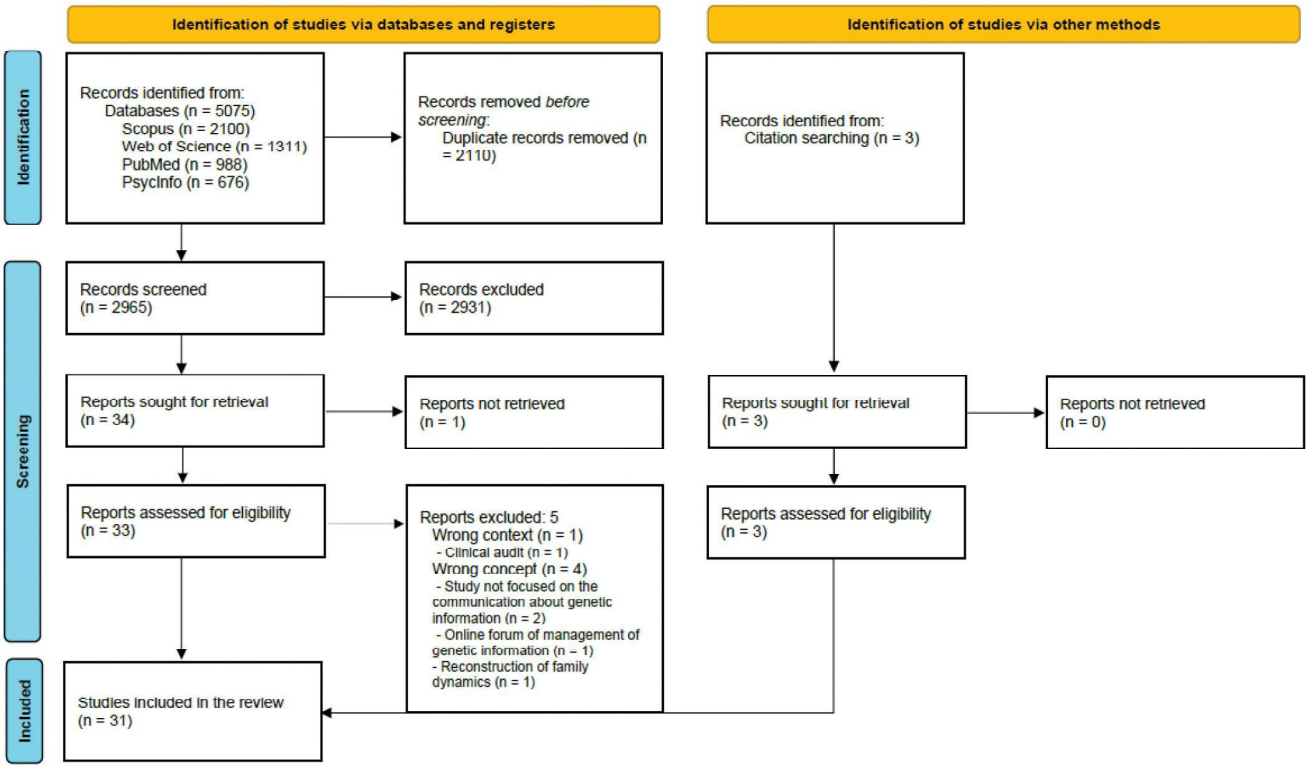


Figure 1. PRISMA flowchart (scoping reviews [ScR]) applied to the current study.

Overview of the included studies

The papers included were published from 1998 to 2023: two (6.45%) from 1998 to 2002, seven (22.58%) from 2003 to 2007, eight (25.81%) from 2008 to 2012, three (9.68%) from 2013 to 2017, and eleven (35.48%) from 2018 to 2023. Considering the authors' country of affiliation, the studies originated from the United States of America (USA)($n = 9$; 29.03%), the United Kingdom ($n = 8$; 25.81%), Portugal ($n = 4$; 12.90%), France ($n = 3$; 9.68%), Canada ($n = 2$; 6.45%), Norway ($n = 1$; 3.23%), Australia ($n = 1$; 3.23%), New Zealand ($n = 1$; 3.23%), and Belgium ($n = 1$; 3.23%). One study was conducted both in Canada, Denmark, and the USA ($n = 1$; 3.23%). All studies were written in English, except one, which was in French. All studies were retrospective, with the large majority employing a qualitative approach ($n = 26$; 83.87%), mainly through semi-structured interviews, although analysis of medical records/clinical notes has also been utilized. Three studies (9.68%) utilized mixed-method approaches and two used quantitative methods (6.45%). The quantitative studies employed statistical analysis methods to examine the data. Qualitative methods used for data analysis in the qualitative studies included grounded theory ($n = 10$; 32.26%), thematic analysis ($n = 6$; 19.35%), interpretative phenomenological analysis ($n = 4$; 12.90%), critical incidents technique ($n = 2$; 6.45%), or were not specified ($n = 2$; 6.45%). A slight majority of the studies did not specify the theoretical frameworks used ($n = 16$; 51.61%), while the remaining studies reported using Family System Theory ($n = 5$; 16.13%), Interpretive Theory of Culture ($n = 2$; 6.45%), Symbolic Interaction Theory ($n = 1$; 3.23%), Family Management Style Framework (FMSF) ($n = 1$; 3.23%), or other frameworks (e.g., descriptive [general] phenomenological approach, broader sociological or psychosocial frameworks, or interdisciplinary theoretical frameworks [$n = 7$; 22.58%]).

The sample size ranged from 8 (Holt, 2006) to 562 participants (Pierron et al., 2023). In 14 cases, the same first author was responsible for more than one publication; four of these articles used the same sample (Etchegary, 2006; Etchegary & Fowler, 2008; Oliveira et al., 2020, 2022). Most of the studies comprised multiple categories of participants (e.g., at risk, pre-symptomatic carriers, non-carriers, and/or non-biological family members) ($n = 26$; 83.87%). The remaining studies included only a) at-risk individuals untested or with unknown test results/risk status ($n = 2$; 6.45%), b) pre-symptomatic carriers ($n = 1$; 3.23%), c) affected ($n = 1$; 3.23%), and d) non-biological family members ($n = 1$; 3.23%). In addition, slightly more than half of the studies ($n = 16$; 51.61%) included multiple family members, such as parents and offspring, siblings, partners, and in-laws. Information regarding the family characteristics of participants was not specified in the remaining studies ($n = 15$; 48.39%). Five articles (16.13%) reported on the ethnicity of the participants, including White, Black, Afro-Caribbean, Asian or Middle East origins, Hispanic, Māori, and "Pākehā" (Forrest et al., 2003; Klitzman & Sweeney, 2011; Metcalfe et al., 2011; Plumridge et al., 2011; Taylor et al., 2019).

Regarding the types of LONDs included in the reviewed studies, the majority focused exclusively on HD ($n = 16$; 51.61%). One study addressed HD and other LONDs such as

Table 2. Summary of the included studies.

Author; year	Country of origin; language	Study aim(s)	Participants (sample size, disease status, sociodemographic and family characteristics)	Methods (study design and approach)	Inherited genetic condition	Main findings about the communication of genetic information
Binedell et al. (1998)	UK; English	To describe the experiences of individuals at risk for HD about their understanding of HD, living at risk, the meaning of a test result, decisions concerning predictive testing and perceptions of the genetics services	*N* = 54 At-risk individuals untested (non-requesters *n* = 32) or with unknown test results/risk status (requesters = 22) Requesters: 10 female, 12 male *M* age = 45.5 years One participant per family (22 families) Non-requesters: 21 female, 11 male *M* age = 42.12 years Multiple participants per family (22 families) in the non-requesters group – kinship not specified	Mixed methods [Quantitative (questionnaires) and qualitative (semi-structured interview)]; retrospective study Statistical analysis; qualitative data analysis methods not specified; Theoretical framework not specified	Huntington disease (HD)	– 31% of the non-requesters mentioned the lack of communication regarding HD and predictive testing; – Lack of communication may contribute to non-requesters' lower levels of knowledge about the test and their reluctance to undergo testing; – A supportive family environment in which genetic issues are discussed may be important in decisions concerning testing; – The decision to undergo testing may depend on the family context.
Delliaux et al. (2008)	France; French	To describe how information about HD is communicated within families and to better understand the transmission of information	*N* = 105 Affected individuals 57 male, 48 female Age range 21 – 81 years (*M* age = 49.8) Detailed information on participant's family characteristics not specified	Mixed methods [Qualitative (semi-structured interview, medical records) quantitative [questionnaire]); retrospective study Data analysis methods not specified Theoretical framework not specified	Huntington disease (HD)	– The decision to disclose genetic risk to relatives was influenced by previous knowledge about the disease in the family context; – The majority of patients wish to inform their children about the disease (80% in cases where the disease was already known in the family, and 92% in cases where there were no family antecedents); – One participant in the "poorly known" disease category chose to hide the existence of HD from their children; – The desire to protect children was mentioned as a motivation to hide communication.
Dimond et al. (2022)	UK; English	To explore how the family is implicated in the patient experience of genetic testing	*N* = 17 Pre-symptomatic carriers and non-carriers Gender and age not specified Detailed information on participant's family characteristics not specified	Qualitative (semi-structured interview); retrospective study Thematic analysis Interdisciplinary theoretical framework (sociology, science and technology studies, anthropology and family studies)	Huntington disease (HD) Hereditary breast and ovarian cancer (HBOC)	– Disclosure was often made by the non-affected parent; – Family members work at gatekeeping, making decisions on whether, when, and how to communicate risk information; – Many of the participants were informed as an adult; – For many participants, the negative test result enabled the communication of results; they acknowledged a reluctance to discuss the disease and the test before knowing the results, and in the case of a positive result they might not have discussed it at all.

(*Continued*)

Table 2. (Continued).

Author; year	Country of origin; language	Study aim(s)	Participants (sample size, disease status, sociodemographic and family characteristics)	Methods (study design and approach)	Inherited genetic condition	Main findings about the communication of genetic information
Etchegary (2006)	Canada; English	To explore how and when participants discovered the family history of HD and to document any differences in pathways to discovery	*N* = 24 At-risk individuals untested or with unknown test results/risk status, affected, pre-symptomatic carriers, non-carriers, non-biological family members 18 female, 8 male Age range 21–73 years (*M* age = 46.2) Multiple participants per family (10 families: kinship not specified)	Qualitative (semi-structured interview); retrospective study Interpretative Phenomenological Analysis Theoretical framework not specified	Huntington disease (HD)	– Initial discovery of HD categorized as (1) "something is wrong", (2) "out of the blue", (3) "knowing, but dismissing," and (4) "growing up with HD;" – Disclosure depended on prior knowledge of HD, the temporal context (adulthood or childhood) and geographical context (proximity or distance), the process of family awareness (immediate or gradual), and the process of realizing the implications for self (immediate or gradual).
Etchegary and Fowler (2008)	Canada; English	To understand how perceptions of responsibility are experienced in test decisions, future planning, and caregiving	*N* = 24 At-risk individuals untested or with unknown test results/risk status, affected, pre-symptomatic carriers, non-carriers, non-biological family members 18 female, 8 male Age range 21–73 years (*M* age = 46.2) Multiple participants per family (10 families: kinship not specified)	Qualitative (semi-structured interview); retrospective study Interpretative Phenomenological Analysis Theoretical framework not specified	Huntington disease (HD)	– Genetic responsibility included communicating with family members, e.g., providing knowledge for future generations, partners and extended family members; – There was a perception of responsibility to protect young children from painful information.
Forrest et al. (2003)	UK; English	To explore and synthesize the factors which influence whether patients share information with relatives and with whom the information is shared	*N* = 37 At-risk individuals untested, affected, pre-symptomatic carriers and non-carriers, non-biological family members *n* HD = 16, *n* HBOC = 21 HD group: 8 female, 8 male *M* age = 46.8 years Detailed information on participant's family characteristics not specified	Qualitative (semi-structured interview); retrospective study Grounded Theory Broader sociological framework	Huntington disease (HD) Hereditary breast and ovarian cancer (HBOC)	– Parents and women are seen as having the primary responsibility for sharing information with relatives; – Parents usually tell children in first key decisions or when they feel that they are old enough to understand; – People first make sense of their own risk before considering whether and what to tell their family members; – Communication is influenced by prior familial and cultural factors and individuals' responses; – "Pragmatics" respondents tended to talk in active and practical terms while "prevaricators" looked for the opportunity within the normal course of family life; – Initial discovery of HD: growing up at risk, finding out unexpectedly when a relative was diagnosed, knowing something was wrong but not what, and knowing about the condition but not of personal risk; – Reasons not to tell relatives are the desire to protect them, and difficulties in overcoming preexisting conflicts or rifts.

(*Continued*)

Table 2. (Continued).

Author; year	Country of origin; language	Study aim(s)	Participants (sample size, disease status, sociodemographic and family characteristics)	Methods (study design and approach)	Inherited genetic condition	Main findings about the communication of genetic information
Hamilton et al. (2005)	USA, Canada, Denmark; English	To describe the experiences of disclosing the results of genetic testing to relatives	*N* = 29 At-risk individuals untested, pre-symptomatic carriers and non-carriers *n* HD = 10, *n* HBOC = 14 Gender and age not specified Detailed information on participant's family characteristics not specified	Qualitative (semi-structured interview); retrospective study Grounded Theory Theoretical framework not specified	Huntington disease (HD) Hereditary breast and ovarian cancer (HBOC)	– Selective disclosure: involves deciding what information to share with certain family members, which extends beyond test results to include risk, details of the condition, and the implications of the results; – The timing of disclosure was related to the readiness of both the teller and the receiver; – Participants considered the potential impact of disclosure on relatives by assessing their vulnerability and receptivity, then selectively chose which information to share and with whom; – Most of the HD participants did not disclosed information beyond the test result unless asked by the sibling to do so.
Holt (2006)	USA; English	To investigate the process of disclosure of genetic risk status to children, especially in the context of predictive genetic testing	*N* = 8(2 families) At-risk individuals untested, affected, and non-biological family members Multiple family members per family Family 1: 3 female, 1 male; father symptomatic (age 47), mother not at risk (age 45), two daughters (age 19 and 16) at 50% risk Family 2: 2 female, 2 male; father symptomatic (age 49), mother not at risk (age 47), son (age 21), and daughter (age 19) at 50% risk	Qualitative (semi-structured interview); retrospective study Interpretative Phenomenological Analysis Theoretical framework not specified	Huntington disease (HD)	– Family 1: deliberate decision to disclose information to children at a very young age, allowing genetic testing to provide knowledge and enabling the family to act together against the condition; – Family 2: deliberate decision not to disclose to children until the affected member became symptomatic and received a definitive diagnosis of HD; – Non-disclosure as means to protect the children emotionally and psychologically and maintain their hope of not having the disease; – Anticipation of anxiety was another inhibitor of family communication; – All of the now adult children expressed a preference to know their genetic status and family history early in life and open/supportive style of communication regarding the condition; – Family tensions and rifts may hinder family communication with first-degree relatives.
Keenan et al. (2005)	UK; English	To explore the barriers and facilitators in family communication about genetic risk	*N* = 56 At-risk individuals untested, affected, pre-symptomatic carriers, non-carriers, and non-biological family members *n* HD = 29, *n* HBOC = 27 HD group: 16 patients who attended genetic counseling, 12 partners, and one other relative (not specified) Patients: 8 female, 8 male Age range 26–65 (*M* age = 46.8 years) Multiple participants per family: patients and their partners/ spouses	Qualitative (semi-structured interview); retrospective study Grounded Theory Sociological framework about contemporary family relations	Huntington disease (HD) Hereditary breast and ovarian cancer (HBOC)	– Parents held the primary responsibility for sharing information with children; – Women are primarily responsible of finding out about genetic risk and sharing information within the family – Participants have different notions and beliefs on who counts as "family" and about their responsibilities and obligations toward different family members; – Participants in general first tell first-degree relatives; telling second and third-degree relatives depends on emotional closeness or contact; – Family communication depends on whom individuals feel obligated to inform, which is influenced by the characteristics of the condition (e.g., available treatment, mode of inheritance)

(*Continued*)

Table 2. (Continued).

Author; year	Country of origin; language	Study aim(s)	Participants (sample size, disease status, sociodemographic and family characteristics)	Methods (study design and approach)	Inherited genetic condition	Main findings about the communication of genetic information
Keenan et al. (2009)	UK; English	To describe the experiences of young people who live in a family affected by HD and their experiences of disclosure	*N* = 33 At-risk individuals untested or with unknown test results, affected, pre-symptomatic carriers and non-carriers 21 female, 12 male *M* age = 20.5 years 1 affected, 26 at risk, 1 carrier, 5 non-carrier Detailed information on participant's family characteristics not specified	Qualitative (semi-structured interview); retrospective study Thematic Analysis Broader sociological framework	Huntington disease (HD)	– Four types of disclosure: (1) "having always been told" (the majority by a female relative), (2) "gradually told", (3) "HD was kept a secret by parents", (4) "HD as a new diagnosis;" – The majority of participants were told in childhood; – Participants whose parents had chosen to keep the condition a secret, were told about HD in late adolescence or adulthood.
Keenan et al. (2013)	UK; English	To describe the experiences of partners in finding out about HD and any impact on couples' marriages/ relationships	*N* = 39 Non-biological family members 14 female, 25 male Age range 25 – 67 years (*M* age = 42) Detailed information on participant's family characteristics not specified	Qualitative (semi-structured interview); retrospective study Thematic Analysis Broader sociological framework	Huntington disease (HD)	– Four types of disclosure: (1) "marital secrets", (2) "alerting but not informing", (3) "knowing and seeing," and (4) "marital ignorance;" – Most participants reported that they had been told about the risk by their partner, and ten described being told by e.g., partner's sibling, aunt/uncle, general practitioner/counselor; – There were participants who wanted to know more about the disease but their partner blocked the process; – Some participants described a more open style of disclosure, which may result in a more detailed awareness of the partner's risk and the impact of the condition
Kjoelaas et al. (2022)	Norway; English	To explore the experiences of both caregivers and offspring on talking about HD throughout childhood	*N* = 50 At-risk individuals with unknown test results/risk status and non-biological family members *n* caregivers = 14, *n* offspring = 36 Offspring: 26 female, 10 male Age range 13–65 years (*M* age = 36.6); Caregivers: 11 female, 3 male Age range 42 – 67 years (*M* age = 54.9) Detailed information on participant's family characteristics not specified	Qualitative (semi-structured interview); retrospective study Thematic Analysis Theoretical framework not specified	Huntington disease (HD)	– Caregivers: described concerns on when offspring should be told about HD, given their maturity or adaptability, while also considering their own needs and that of the offspring and timing of disclosure; reported difficulties regarding what to say and how often they need to talk with children; they weigh the consequences of being honest vs cautious and not providing enough information or misleading, inaccurate information; – Offspring: highlighted the importance of being told in planned events rather than spontaneously; wanted to learn about the disease as early as possible but hesitate over whether all information should be disclosed, how often it should be talked daily

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Author; year	Country of origin; language	Study aim(s)	Participants (sample size, disease status, sociodemographic and family characteristics)	Methods (study design and approach)	Inherited genetic condition	Main findings about the communication of genetic information
Klitzman et al. (2007)	USA; English	To examine what, how, and when to disclose genetic information to family members	*N* = 21 At-risk individuals untested, affected, pre-symptomatic carriers and non-carriers 8 female, 13 male *M* age not specified Detailed information on participant's family characteristics not specified	Qualitative (semi-structured interview); retrospective study Grounded Theory Interpretive Theory of Culture	Huntington disease (HD)	– Participants struggled with questions of what to tell, e.g., having symptoms or the mutation, undergoing genetic counseling and testing, or disclosing the results; – Coded or partial information may be shared; – Disclosure can occur gradually or abruptly; – Discussions with the children can be indirect or non-verbal, and partial and incomplete information can lead to misunderstanding; – Disclosure often occurs due to the perceived responsibility to foster the health of relatives, pursue medical evaluation and family planning, and avoid further transmission of the genetic alteration; – Non-disclosure was justified to avoid burdening their offspring; distance and closedness and strained and estranged relationships were identified as a barrier; – Psychiatric symptoms may impede disclosure, which can lead to stigma and discrimination; – When to disclose depended on the family life cycle and the medical course of the affected members, age and maturity of the offspring; – Disclosure of diagnosis depends on previous family communication style and prior knowledge of the condition; – Disclosing to family members may depend on the type of relationship (immediate vs. distant family; emotionally close vs distant)

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Author; year	Country of origin; language	Study aim(s)	Participants (sample size, disease status, sociodemographic and family characteristics)	Methods (study design and approach)	Inherited genetic condition	Main findings about the communication of genetic information
Klitzman and Sweeney (2011)	USA; English	To examine how individuals broach the topic of genetic risk information in dating situations; whether they discuss genetic information, and if so, when, to what extent, how, and with whom	*N* = 64 At-risk individuals untested, affected, pre-symptomatic carriers and non-carriers *n* BRCA = 32; *n* HD = 21; *n* Alpha = 11 HD group: 9 female, 12 male Age range 21–80 years (*M* age not specified) Detailed information on participant's family characteristics not specified	Qualitative (semi-structured interview); retrospective study Grounded Theory Interpretive Theory of Culture	Huntington disease (HD) Hereditary breast and ovarian cancer (HBOC) Alpha-1 antitrypsin deficiency	– Decisions were made based on medical factors (e.g., type of condition, family history, symptoms, treatability, testing, and results), psychological (e.g., fears and past experiences of rejection, desires for children), and social (e.g., the prospective partner's expectations and desires concerning the relationship and children, compassion, and relationship's length and nature); – Reasons to tell: ethical obligation, impossibility of concealing the disease within the family or from themselves, children would be affected, and avoidance of dishonesty, concealment, or perceived lack of trust; – Reasons not to tell: fear of rejection (breakups); –Difficulties on how much detail to provide arise concerning prognosis and reproductive implications, but also the course of the illness, so that partial or incomplete information may be provided; –Information may be framed and/or delivered in person or otherwise (e.g., by e-mail); –Participants also expressed concerns about the timing of disclosure, which may occur when the relationship is "close enough," if symptoms became visible, when asked directly, after a certain length of time, or if a relationship breakup occurs.
Mendes et al. (2017)	Portugal; English	To investigate experiences of stigmatization by those affected with or at risk of Familial Amyloid Polyneuropathy	*N* = 11 Affected individuals and pre-symptomatic carriers 6 female, 5 male Age range 25–49 years (*M* age = 40) Detailed information on participant's family characteristics not specified	Qualitative (semi-structured interview); retrospective study Interpretative Phenomenological Analysis Broad psychosocial framework	Familial Amyloid Polyneuropathy	– Discussing the condition was mainly kept within the family; – Affected families often feel they can only discuss the condition within the family, as they tend to perceive the broader community to be stigmatizing

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Table 2. (Continued).

Author; year	Country of origin; language	Study aim(s)	Participants (sample size, disease status, sociodemographic and family characteristics)	Methods (study design and approach)	Inherited genetic condition	Main findings about the communication of genetic information
Metcalfe et al. (2011)	UK; English	To explore the communication of genetic risk between parents and children and the factors involved	*N* = 79 At-risk individuals untested or with unknown results/risk status, affected, pre-symptomatic carriers, non-carriers, and non-biological family members Families: *n* NF = 4, *n* HD = 7, *n* HBO = 6, *n* FAP = 6, *n* DMD = 6, *n* CF = 4 Age range parents 30–65+ years Age range children 8–18+ years Multiple participants per family (33 families): children, siblings, parents	Qualitative (semi-structured interview); retrospective study Grounded Theory Family System Theory, Symbolic Interaction Theory	Huntington disease (HD) Neurofibromatosis (NF) Hemoglobinopathies (HBO) Familial Adenomatous Polyposis (FAP) Duchene muscular dystrophy (DMD) Cystic fibrosis (CF)	– Mothers are usually the key person who share genetic information; – Children and young people preferred less formal discussions, in which they were doing other things alongside their parents; – When parents shared information about the genetic condition and its risk as an ongoing process throughout childhood, greater insight and coping with the condition and increased knowledge of associated risk were reported [unclear whether this result is specific to LONDs].
Oliveira et al. (2017)	Portugal; English	To understand the role of the older generation in families affected by Familial Amyloid Polyneuropathy	*N* = 18 At-risk individuals untested, affected, pre-symptomatic carriers, non-carriers, and non-biological family members 10 female, 8 male Age range 18–65 years (*M* age = 42) 7 participants did not have family relationships within the sample (one participant per family), and 11 participants were members of 4 families (Multiple participants per family – brothers, sisters, sisters-in-law, cousins, sons, mothers)	Qualitative (semi-structured interview); retrospective study Critical Incidents Technique Theoretical framework not specified	Familial Amyloid Polyneuropathy	– Older generations may play a role in informing, and supporting family members; – The intergenerational flow of information involves primarily women, occurring from mother to daughter, and from older affected individuals to young pre-symptomatic carriers; – Informing occurs when the older relatives share what they know about the condition (e.g., history in the family, characteristics of the disease and old myths); – Silencing occurs when the older individual does not talk about the condition, which may be due to the associated suffering when remembering disease-related experiences
Oliveira et al. (2020)	Portugal; English	To explore how families manage information about HD, from a transgenerational perspective	*N* = 10 Pre-symptomatic carriers, non-carriers, and non-biological family members 8 female, 2 male Age range 28–72 years (*M* age = 45.9) Multiple participants per family (7 families): mothers, daughters, uncles, nephews, sisters-in-law, spouses, ex-spouses	Qualitative (semi-structured interview); retrospective study Grounded Theory Theoretical framework not specified	Huntington disease (HD)	– Family members manage genetic information through (1) closedness, i.e., denying the disease or preferring not to know their own or their relatives' genetic status, avoiding talking about the disease, due to fear of rejection and protecting others, and (2) openness, i.e., accepting the disease, without expressing shame or embarrassment, searching for information, becoming active in the community and being open about sharing this information with other relatives and the community – Some participants reported shame and stigma as leading to keep HD a secret

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Table 2. (Continued).

Author; year	Country of origin; language	Study aim(s)	Participants (sample size, disease status, sociodemographic and family characteristics)	Methods (study design and approach)	Inherited genetic condition	Main findings about the communication of genetic information
Oliveira et al. (2022)	Portugal; English	To explore the intergenerational flow of health-related roles from older to younger generations	*N* = 10 Pre-symptomatic carriers and non-carriers, and non-biological family members 8 female, 2 male Age range 28–72 years (*M* age = 45.9) Multiple participants per family (7 families): mothers, daughters, uncles, nephews, sisters-in-law, spouses, ex-spouses	Qualitative (semi-structured interview); retrospective study Critical Incidents Technique Family Systems Theory	Huntington disease (HD)	– Older generations play two main roles in families with HD: "shaping awareness" and "influencing management;" – Promoting awareness occurs through sharing testimonies about the condition, or when a younger relative learns about the condition through witnessing it from an older relative, or by providing information to younger relatives; hampering awareness manifested through silence and denial; – The role of shaping awareness was performed mostly by mothers toward daughters, and family members over 60 years.
Penziner et al. (2008)	USA; English	To identify perceptions about discrimination in persons who have received predictive testing for HD	*N* = 15 Pre-symptomatic carriers 11 female, 4 male Age range 22–62 years (*M* age not specified) Detailed information on participant's family characteristics not specified	Qualitative (semi-structured interview); retrospective study Content Analysis Theoretical framework not specified	Huntington disease (HD)	– Most participants (87%) disclosed their test results to their family and social network; – Patterns of disclosure varied from being cautious or open to sharing test results; – Two participants withheld their testing experience from certain relatives, while selectively sharing the test results with other relatives who they believed to be more supportive.
Pierron et al. (2021)	France; English	To compare how information concerning genetic risk in HD is disclosed with families two decades apart	*N* = 148 in 2000 97 female, 51 male and *N* = 295 in 2019 195 female, 100 male At-risk individuals with unknown test results/risk status, affected, pre-symptomatic carriers, non-carriers, and non-biological family members *M* age = 32.2 (in 2000) and 29.0 (in 2019) Detailed information on participant's family characteristics not specified	Quantitative (questionnaires); retrospective study Statistical analysis Theoretical framework not specified	Huntington disease (HD)	– Parents had primarily provided information on the transmission of risk; – In 2000 (*n* = 148), 26 participants reported being informed by parents, and 12 by other relatives, 50 responded that the age to inform children should be <18 years; – In 2019 (*n* = 295): 83 reported being informed by parents, 30 by other family members, 123 responded that the age to inform children should be <18 years; – The motivations to recommend testing to relatives included the "right to know" about their status and non-transmission to future offspring, to anticipate and plan for the future, and to potentially benefit from treatment; – The motivations not to recommend testing to relatives included concern for individual choice, the potential negative psychological impact, and the absence of a cure.

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Author; year	Country of origin; language	Study aim(s)	Participants (sample size, disease status, sociodemographic and family characteristics)	Methods (study design and approach)	Inherited genetic condition	Main findings about the communication of genetic information
Pierron et al. (2023)	France; English	To investigate and compare the perceptions of reproductive choices in several autosomal dominant neurological diseases	*N* = 562 At-risk individuals with unknown test results/risk status, affected, pre-symptomatic carriers and non-carriers, non-biological family members 365 female, 197 male *M* age = 46.3 years *n* HD = 307; *n* SCA = 114; *n* MD1 = 82; *n* ALS/FTD = 59 Detailed information on participant's family characteristics not specified	Quantitative (questionnaires); retrospective study Statistical analysis Theoretical framework not specified	Huntington disease (HD) Spinocerebellar ataxia (SCA) Familial Creutzfeldt-Jakob disease (CJD) Amyotrophic lateral sclerosis/frontotemporal dementia (ALS/FTD) Steinert myotonic dystrophy (MD1)	– 53.3% of participants mentioned prenatal/preimplantation diagnosis as a motivation to inform relatives, which was not significantly different between conditions.
Plumridge et al. (2011)	UK; English	To explore parents' communication of genetic risk information with siblings of children affected by an inherited genetic condition	*N* = 96 Affected individuals, pre-symptomatic carriers, non-carriers, and unspecified risk status (parents) *n* parents = 52, 34 female, 18 male; *n* children = 33 (<18 years) and *n* adult children = 11 (18+ years); 24 female, 20 male Genetic condition among families: *n* NF = 4, *n* HD = 7, *n* HBO = 6, *n* FAP = 6, *n* DMD = 6, *n* CF = 4 Multiple participants per family (33 families): parents and children/young people, siblings	Qualitative (semi-structured interview); retrospective study Grounded Theory Family Systems Theory	Huntington disease (HD) Neurofibromatosis (NF) Hemoglobinopathies (HBO) Familial Adenomatous Polyposis (FAP) Duchene muscular dystrophy (DMD) Cystic fibrosis (CF)	– Affected and unaffected children consistently said that they wanted to receive information from a young age and be able to discuss it within their families throughout childhood.
Poulton et al. (2023)	Australia; English	To investigate how individuals experience family communication about genetic risk for YOD	*N* = 9 At-risk individuals with unknown risk status, and non-biological family members *n* AD = 7, *n* FTD = 2 and unspecified type = 1 Gender not specified Age range 20–79 years (*M* age not specified) Detailed information on participant's family characteristics not specified	Qualitative (semi-structured interview); retrospective study Thematic Analysis Descriptive (general) phenomenological approach	Young onset dementia (YOD)	– Communication about genetic risk may be affected by barriers such as preexisting family tension, and/or disconnection, geographical distance, and avoidant coping strategies; – Most participants communicated genetic risk with immediate adult family members before evaluating testing for the patient; – Conversations with children, adult family members who were unreceptive to initial conversations, and extended family depend on the identification of a genetic component/test results; – Autonomy of the family member emerged as a motivation for communicating genetic risk; in many cases, communication occurred to facilitate informed, autonomous decision-making for the next generation.

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Author; year	Country of origin; language	Study aim(s)	Participants (sample size, disease status, sociodemographic and family characteristics)	Methods (study design and approach)	Inherited genetic condition	Main findings about the communication of genetic information
Sobel and Cowan (2000)	USA; English	To explore the impact of genetic testing from the families' perspective	*N* = 55 Pre-symptomatic carriers, and non-carriers Age (at testing) range 21–67 years Gender not specified Multiple participants per family (18 families): kindship not specified	Qualitative (semi-structured interview); retrospective study Qualitative analysis not specified Family Systems Theory	Huntington disease (HD)	– HD was either misdiagnosed or known to exist but kept a secret in these families' parents' generation; – Nine families (50%) stated that testing did not change their pattern of communication in the family about the condition in the nuclear or extended family; the remaining families report changes in their communication patterns; – In five families, all members of the family of origin, the nuclear family, and the extended family know of the test results but there was no discussion about it; – One family reported that it has always had open and direct communication about the condition; – For some families the positive test result, the communication was restricted; – Some members used testing as an opportunity to talk, while others maintained previous patterns of silence; – Sometimes information about the condition was withheld out of a sense of protection or misinformation.
Stuttgen et al. (2020)	USA; English	To examine if and when individuals communicated HD risk, the decision to pursue testing, and test results to family members and others before, during, and up to three decades after testing	*N* = 39 Pre-symptomatic carriers, and non-carriers 20 female, 19 male *M* age = 59.6 years Detailed information on participant's family characteristics not specified	Qualitative (semi-structured interview; clinical notes); retrospective study Grounded Theory Theoretical framework not specified	Huntington disease (HD)	– Communication within families depended on the type of family relationship (e.g., significant others, children, extended family); – All participants with a significant other at the time of testing communicated genetic risk, to pursue testing, and test result; – Individuals who knew their risk status before meeting their partner, communicated information while dating; when they learned about their risk after, partners were informed very soon, if not immediately; – All individuals with children informed them of the test result, irrespective of the genetic status; the timing and frequency of communication depended on the age of children and the proband's mutation status; – Reasons for sharing results: responsibility to inform other family members about risk and testing, obtaining emotional support, preparing others for symptom onset, and relieving worry; – Reasons to not share results: denial and/or lack of communication about the condition in the family, lack of support from relatives throughout testing, preexisting conflicts in the family, and guilt after receiving a normal-repeat result; – Poor family communication was associated with a more serious family trauma (i.e., verbal or physical abuse)

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Author; year	Country of origin; language	Study aim(s)	Participants (sample size, disease status, sociodemographic and family characteristics)	Methods (study design and approach)	Inherited genetic condition	Main findings about the communication of genetic information
Stuttgen et al. (2021)	USA; English	To understand how parents and minors view genetic risk/ status and how it is communicated	*N* = 85 At-risk individuals with unknown risk status, affected, and non-biological family members 22 female children, 12 male children 28 female parents, 4 male parents Parents *n* HCs = 17, *n* HD = 15 *n* GC = 19 Age not specified Minors at risk for *n* HCs = 19, *n* HD = 15 Age range of minors 15–17 years Multiple participants per family: parents, children	Qualitative (semi-structured interview); retrospective study Content Analysis Theoretical framework not specified	Huntington disease (HD) Hereditary cancer (HC)	– All mature minors were happy their parent had disclosed genetic risk information; – All mature minors felt that parents should have conversations with minors about the risk several times over months or years; – Almost all mature minors reported that the parent should honestly communicate the information [unclear whether the result is specific to LONDs]; – Nearly all parents and mature minors felt that the parent should first disclose [unclear whether the result is specific to LONDs]; – Parents reported wanting to protect minors from the information.
Taylor et al. (2019)	New Zealand; English	To explore the experience of families about the DM1 diagnosis	*N* = 17 Affected individuals, and non-biological family members 11 female, 6 male Age range 25–79 years (*M* age = 54.9) Multiple participants per family: mothers, daughters, sons, spouses	Qualitative (semi-structured interview); retrospective study Thematic Analysis Family System Theory	Type 1 myotonic dystrophy (DM1)	– Some participants described their experiences of open communication; others described that conversations were avoided; – Most participants reported that women were the communication key person; – Motivations to inform relatives: making informed reproductive and medical decisions; – Barriers to communication: disconnection, estrangement, conflict, adoption, and geographical distance.
Van Riper (2005)	USA; English	To explore the family experience of genetic testing	*N* = 6 Non-carriers and non-biological family members Family with HD (*n* = 3) and HBOC (*n* = 3) HD: 2 female, 1 male Age not specified Multiple participants per family: daughters and father/husband of an affected women	Qualitative (semi-structured interview); retrospective study Qualitative data analysis not specified Family Management Style Framework (FMSF)	Huntington disease (HD) Hereditary breast and ovarian cance (HBOC)	– One participant reported difficulties telling at-risk family members the results of genetic testing and also marital problems after finding out the results; – This participant tried to encourage and facilitate open communication about the condition in both her immediate and extended family.
Wain et al. (2009)	USA; English	To understand the experience of adults who have a sibling with early-onset AD, their sense of risk and the impact of their sibling's illness on life decision	*N* = 25 At-risk individuals untested or with unknown test results/risk status 15 female, 10 male Age range (*M* age = 56.8 years) Multiple participants per family (17 families): all siblings of people diagnosed	Mixed methods [Qualitative (semi-structured interview) and quantitative (questionnaires)]; retrospective study Statistical analysis; Grounded Theory Theoretical framework not specified	Alzheimer's disease (AD)	– Nine participants reported that there is, or was, some difficulty talking about the condition in the family.

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Author; year	Country of origin; language	Study aim(s)	Participants (sample size, disease status, sociodemographic and family characteristics)	Methods (study design and approach)	Inherited genetic condition	Main findings about the communication of genetic information
Wauters and Van Hoyweghen (2021)	Belgium; English	To investigate worry about genetic discrimination among individuals with a predisposition to HD and the coping strategies used	*N* = 12 At-risk individuals untested or with unknown risk status, pre-symptomatic carriers and non-carriers 10 female, 2 male Age range 28–60 years (*M* age = 41) Detailed information on participant's family characteristics not specified	Qualitative (semi-structured interview); retrospective study Grounded Theory Theoretical framework not specified	Huntington disease (HD)	– Some participants persisted in a family pattern of secrecy; – Most participants had been aware of the condition for decades; while some individuals were aware of the impact of HD in relatives, others had perceived that "something was wrong;" – One way of coping with fears of genetic discrimination was persisting in a family pattern of secrecy; another is breaking with the secretiveness of relatives and to raise awareness.

spinocerebellar ataxia (SCA), amyotrophic lateral sclerosis/frontotemporal dementia (ALS/FTD), or Steinert myotonic dystrophy (MD1) (3.23%). Nine studies focused on LONDs and non-LONDs such as hereditary breast and ovarian cancer (HBOC), neurofibromatosis (NF), familial adenomatous polyposis (FAP), Duchenne muscular dystrophy (DMD), hemoglobinopathies (HbO), cystic fibrosis (CF), and others (29.03%). The remaining studies involved exclusively families with familial amyloid polyneuropathy ($n = 2$; 6.45%), Alzheimer's disease (AD) ($n = 1$; 3.23%), type 1 myotonic dystrophy (DM1) ($n = 1$; 3.23%), and young onset dementia (YOD) ($n = 1$; 3.23%). The contexts in which the participants were recruited included clinical services ($n = 7$, 22.58%), patient associations, support groups, or websites ($n = 7$; 22.58%), national or research registers ($n = 1$; 3.23%), and personal contact networks ($n = 1$; 3.23%). In some cases, the recruitment context comprised two or more of the mentioned contexts ($n = 13$; 41.94%). The context of participant recruitment was not specified in two studies (6.45%).

Characteristics of the communication of genetic information in families with LOND

The main categories of the communication of genetic information with family members with LONDs were: (1) from whom to whom: communication transmitters and receivers, (2) what to tell: content and details of communication, (3) how to tell: communication approach, (4) when to tell: timing of communication, (5) barriers and reasons for not communicating, and (6) facilitators and reasons for communicating.

From whom to whom: communication transmitters and receivers

All the studies ($n = 31$) identified the individuals involved in the communication process of genetic information, specifically the transmitters and the receivers of information. Parents (either affected by the condition or not) were frequently described as transmitters of the information to their offspring (Dimond et al., 2022; Forrest et al., 2003; Holt, 2006; Keenan et al., 2009; Kjoelaas et al., 2022; Metcalfe et al., 2011; Pierron et al., 2021; Plumridge et al., 2011; Stuttgen, et al., 2021). Several studies reported that the flow of information primarily involved women (Keenan et al., 2009; Metcalfe et al., 2011; Oliveira et al., 2017, 2022; Taylor et al., 2019). A pivotal person was identified as being responsible for finding out about genetic risk and sharing this information with other family members (e.g., a grandmother) (Keenan et al., 2005). In two studies, older affected mothers were typically acknowledged as the transmitters of information about the disease to pre-symptomatic carriers (Oliveira et al., 2017, 2022).

Transmitters often selected whom to share genetic information with based on their understandings of family, kinship, and type of relationship (i.e., partners, children, and extended family members), as well as their awareness of relatives' testing status and attitudes toward testing (Forrest et al., 2003; Keenan et al., 2005; Klitzman et al., 2007; Stuttgen et al., 2020). First-degree relatives were identified as the main recipients of genetic information. In one study, communication with second- and third-degree relatives was commonly based on the closeness of their relationship or their degree of contact (Keenan et al., 2005).

Selecting whom to tell may also involve assessing the family member's vulnerability and receptivity to the information (Hamilton et al., 2005). The partners of individuals at risk, who are typically not biologically related, may be informed by their partners. If these are reluctant to do it, the partners' own relatives will sometimes do so, such as their parents (the "in-laws"), their siblings, or aunts/uncles (Keenan et al., 2013). Communication also involved other groups, including both minor and adult siblings (Metcalfe et al., 2011; Plumridge et al., 2011; Wain et al., 2009) (cf. *timing of communication*).

What to tell: content and details of communication

Six studies (19.35%) (Hamilton et al., 2005; Kjoelaas et al., 2022; Klitzman & Sweeney, 2011; Klitzman et al., 2007; Oliveira et al., 2017; Stuttgen et al., 2021) addressed the specific content and details of the communication between family members.

The content included information on disease progression and prognosis, symptoms, treatment options, and reproductive implications (Hamilton et al., 2005; Klitzman & Sweeney, 2011; Klitzman et al., 2007). It may also comprise information about the genetic counseling process and testing (Klitzman et al., 2007), and the implications of test results for children (Stuttgen et al., 2021). One study reported that older generations tend to share the family history of the disease with younger family members and its characteristics, and old myths about how the disease was perceived (Oliveira et al., 2017). Other studies addressed the difficulties surrounding what and how much information should be disclosed to family members, which often resulted in incomplete or partial disclosure (Hamilton et al., 2005; Kjoelaas et al., 2022; Klitzman & Sweeney, 2011; Klitzman et al., 2007). One study reported that transmitters may decide whether to disclose coded or only partial information (e.g., that counseling has been initiated or that they underwent genetic testing) (Klitzman et al., 2007).

How to tell: communication approach

Nineteen studies (61.30%) explored how communication occurred in families (Binedell et al., 1998; Dimond et al., 2022; Etchegary, 2006; Forrest et al., 2003; Hamilton et al., 2005; Keenan et al., 2009, 2013; Kjoelaas et al., 2022; Klitzman & Sweeney, 2011; Klitzman et al., 2007; Metcalfe et al., 2011; Oliveira et al., 2020; Penziner et al., 2008; Plumridge et al., 2011; Sobel & Cowan, 2000; Stuttgen et al., 2020 Taylor et al., 2019; Wauters & Van Hoyweghen, 2021). These studies characterized communication as a process that ranges from gradual to abrupt disclosure, with varying degrees of communication openness (Etchegary, 2006; Keenan et al., 2009; Klitzman et al., 2007; Metcalfe et al., 2011, Penziner et al., 2008; Stuttgen et al., 2020; Stuttgen et al. 2021). Some studies identified an approach that involves the realization that "something was wrong," which was previously unknown to the family until a relative started to show symptoms (Etchegary, 2006; Forrest et al., 2003). One study reported that disclosing to young people who are unaware of the IGC or their own risk can be followed by a gradual process of disclosure. This approach involved the initial sharing of limited information, followed by providing more detailed information incrementally over time (Keenan et al., 2009). There were also situations where young people were told early, allowing them to "grow up" aware of their family's medical history and knowing that a family member had the condition (Etchegary, 2006; Forrest et al., 2003; Keenan et al., 2009). One study described that families either opted to "keep it a secret" until late adolescence or adulthood, allowing children to grow up around affected relatives, or they chose to avoid contact with any affected relatives, or remaining completely ignorant of the IGC (Keenan et al., 2009). "Knowing but dismissing" corresponded to the approach where an at-risk individual knows that a (usually) distant family member has the disease but is not aware of personal risk (Etchegary, 2006). Another approach was when the diagnosis was disclosed unexpectedly ("out of the blue") to those without prior knowledge of the condition (Etchegary, 2006; Forrest et al., 2003; Keenan et al., 2009).

In some studies, parents either feared the consequences of being completely honest when discussing the condition with their children or ended up providing insufficient and often misleading information (Binedell et al., 1998; Kjoelaas et al., 2022). Some families may communicate indirectly about the IGC (e.g., by recognizing mutual anxiety or unease) or through non-verbal clues (e.g., placing a booklet about HD on the table) (Klitzman et al., 2007). One study described that some parents engage in less formal discussions, such as sharing a "story" about the condition, explaining how they discovered it and learned to cope with it, while others discussed the IGC during other activities (Metcalfe et al., 2011).

The experiences of partners in finding out about HD were also described. One study identified the following approaches: (i) not being told by the partner until marriage; (ii) being alerted by a partner with no direct experience of HD in the family; (iii) knowing about HD as a result from being informed about a partner's risk but also having experiential knowledge from seeing an affected mother or father-in-law; and (iv) finding out about HD unexpectedly due to partner's lacked knowledge of the HD risk (Keenan et al., 2013). Another study noted that partners often choose to present genetic information in positive terms, e.g., by not solely focusing on medical facts, but rather sharing the information as positively as possible or avoiding negativity (Klitzman & Sweeney, 2011).

Communication about a LOND may occur in a closed manner, characterized by avoiding discussion, denying the condition, or expressing a desire not to be reminded of the genetic risk. Conversely, it can also be more open, involving acceptance and discussions with relatives who have the condition (Oliveira et al., 2020). One study described communicative approaches that included actively conveying information in practical terms and seeking opportunities to disclose it during normal family life activities (e.g., get-togethers) (Forrest et al., 2003).

When to tell: timing of communication

Seventeen studies (54.84%) described the timing of communication of genetic information in families (Dimond et al., 2022; Etchegary, 2006; Etchegary & Fowler, 2008; Forrest et al., 2003; Hamilton et al., 2005; Holt, 2006; Keenan et al., 2005, 2009, 2013; Kjoelaas et al., 2022; Klitzman & Sweeney, 2011; Klitzman et al., 2007; Metcalfe et al., 2011; Plumridge et al., 2011; Pierron et al., 2021; Stuttgen et al., 2020; Stuttgen et

al. 2021). Planning and finding "the right time" for disclosure may be influenced by various factors, including the perceived readiness of both the transmitter and the receiver of the information, the family life cycle, the planning of life decisions, the risk status of the transmitter and the receiver, and the disease progression of affected members (Forrest et al., 2003; Hamilton et al., 2005; Klitzman et al., 2007; Stuttgen et al., 2020; Stuttgen et al. 2021). It was also described that people often prioritized making sense of their risk before deciding on whether and what to tell their relatives (Forrest et al., 2003).

Family disclosure can occur at various stages throughout the family life cycle (Etchegary, 2006). Decisions about when to tell children were influenced by their age as well as the parents' perceptions of their children's maturity, individual needs, and life decisions (e.g., marriage, engagement, or having children) (Etchegary & Fowler, 2008; Forrest et al., 2003; Kjoelaas et al., 2022; Klitzman et al., 2007). Parents reported challenges in deciding whether to inform their children too soon or too late, and whether an early discussion would facilitate adjustment over time, or if waiting until maturity would lead to a better understanding of the situation (Kjoelaas et al., 2022). In families that tended to keep the condition a secret during childhood, children may only be told during their adolescence (Keenan et al., 2009). Children retrospectively described a preference for being informed early and openly about the family's medical history and genetic risk (Holt, 2006; Plumridge et al., 2011). Studies also reported that being gradually informed at a young age may help them to develop a greater understanding and knowledge of the condition, enabling them to accommodate their risk over time (Metcalfe et al., 2011) and to plan for life decisions (Kjoelaas et al., 2022). This gradual approach also helped them to avoid the shock of learning about the condition and its implications all at once (Kjoelaas et al., 2022; Metcalfe et al., 2011).

Communication with partners was described as being influenced by the perceived nature of the relationship and the onset and visibility of symptoms. One study reported that individuals who were aware of their risk before meeting their partner typically disclosed this information while dating. In cases where individuals learned their risk after meeting their partner, they communicated this information very soon, often immediately (Stuttgen et al., 2020). In some studies, individuals disclosed genetic information to their partners only when the relationship showed signs of a serious commitment such as long-term potential, or the likelihood of marriage; in other instances, disclosure occurred only when the partner explicitly asked for it or after the relationship had ended (Keenan et al., 2013; Klitzman & Sweeney, 2011).

Barriers and reasons for not communicating

Twenty-three studies (74.19%) described the reasons for not disclosing genetic information to family members and the barriers encountered in that hindered communication (Delliaux et al., 2008; Dimond et al., 2022; Etchegary & Fowler, 2008; Forrest et al., 2003; Holt, 2006; Keenan et al., 2005, 2009, 2013; Klitzman et al., 2007; Klitzman & Sweeney, 2011; Mendes et al., 2017; Oliveira et al., 2017, 2020; Penziner et al., 2008; Pierron et al., 2021; Plumridge et al., 2011; Poulton et al., 2023; Sobel & Cowan, 2000; Stuttgen, Bollinger, et al., 2020; Stuttgen et al. 2021; Taylor et al., 2019; Wain et al., 2009; Wauters & Van Hoyweghen, 2021). These reasons include communication difficulties arising from limited knowledge about the condition and genetic risk (Sobel & Cowan, 2000). One study reported that some relatives do not feel responsible or obligated to inform at-risk relatives (Keenan et al., 2005). Older affected family members also reported remaining silent about the condition with other family members due to the suffering they experienced (Oliveira et al., 2017).

Additionally, several studies described a desire to shield family members, particularly children, from psychologically distressing information (Delliaux et al., 2008; Etchegary & Fowler, 2008; Forrest et al., 2003; Holt, 2006; Oliveira et al., 2020; Sobel & Cowan, 2000; Stuttgen et al., 2021), to avoid practical discrimination (e.g., difficulties in obtaining insurance) (Forrest et al., 2003), as well as to maintain hope (Holt, 2006). Another cited reason for withholding communication was the absence of a cure for the LOND (Pierron et al., 2021).

Fear of rejection and discrimination by close relatives or partner can also hinder family communication (Klitzman & Sweeney, 2011; Oliveira et al., 2020). Additionally, fear of stigmatization has been described as a barrier for discussing the condition within the family (Klitzman et al., 2007; Mendes et al., 2017; Wain et al., 2009), which in the case of HD, that is specifically related to the stigma associated with psychiatric symptoms (Klitzman et al., 2007). Other barriers to communication included denial of the risk or of the diagnosis within the family (Stuttgen et al., 2020; Wauters & Van Hoyweghen, 2021), as well as serious family trauma (e.g., verbal or physical) (Stuttgen et al., 2020). One study reported that several participants were reluctant to discuss the IGC in the family before receiving their test results. The participants anticipated that knowing a positive test result (i.e., identifying the pathogenetic variant) would have obstructed communication, as they sought to avoid placing a sense of blame on other family members (Dimond et al., 2022). Receiving a negative test result (i.e., absence of the pathogenetic variant) may lead to feelings of guilt toward others in the family which can hinder communication (Stuttgen et al., 2020). Families characterized by blocked, restricted, closed, and cautious communication styles are described as experiencing additional barriers to communicating (Penziner et al., 2008; Plumridge et al., 2011; Sobel & Cowan, 2000). Lack of support and/or conflicts among family members, avoidant coping strategies, and geographical distance also tend to block the dissemination of genetic information within the family (Keenan et al., 2005, 2013; Klitzman et al., 2007; Poulton et al., 2023; Taylor et al., 2019).

Facilitators and reasons for communicating

Twenty-two studies (70.97%) described the reasons for disclosing genetic information to family members and the facilitators involved in that process (Binedell et al., 1998; Dimond et al., 2022; Etchegary & Fowler, 2008; Forrest et al., 2003; Hamilton et al., 2005; Holt, 2006; Keenan et al., 2009, 2013; Klitzman et al., 2007; Klitzman & Sweeney, 2011; Metcalfe et al., 2011; Oliveira et al., 2017, 2020; Penziner et al., 2008; Pierron et al., 2021, 2023; Plumridge et al., 2011; Poulton et al., 2023; Sobel & Cowan, 2000; Stuttgen et al., 2020; Taylor et al., 2019; Wauters & Van Hoyweghen, 2021). Feelings of

responsibility or moral obligation to inform family members about their genetic risk and promote their health and autonomy were frequently reported (Binedell et al., 1998; Etchegary & Fowler, 2008; Forrest et al., 2003; Hamilton et al., 2005; Klitzman & Sweeney, 2011; Klitzman et al., 2007; Oliveira et al., 2017; Poulton et al., 2023; Stuttgen et al., 2020; Taylor et al., 2019). Obtaining emotional support from family members while preparing them for symptom onset was also described as a facilitator, along with relief from worry following negative test results for HD (Stuttgen et al., 2020). Other motivations included facilitating family planning, avoiding the transmission of the condition to offspring, and making more informed decisions about reproduction (Klitzman et al., 2007; Pierron et al., 2021, 2023). For partners, sharing genetic information was associated with avoiding dishonesty or mistrust within the relationship, the impossibility of concealing the diagnosis and symptoms, and preventing burdensome obligations to current and future partners (Etchegary & Fowler, 2008; Klitzman & Sweeney, 2011). Additionally, an open or honest communication style within the family, along with a supportive and cohesive family environment, was identified as recurring facilitators of sharing genetic information (Keenan et al., 2009, 2013; Metcalfe et al., 2011; Penziner et al., 2008; Plumridge et al., 2011; Sobel & Cowan, 2000; Stuttgen et al., 2020).

Discussion

This scoping review aimed to map the research evidence on the communication of genetic information in families with LONDs. To our knowledge, this is the first review that specifically addresses family communication within this set of conditions. Understanding this process is timely because, as genetic and genomic testing become widespread, individuals and families will increasingly face complex decisions about sharing genetic information (McNeill, 2022). This review highlights that family communication is a deliberative multi-step process that extends beyond the moment of disclosure. The extent to which individuals communicate information about the IGC with family members, including when and how to do so, who is informed, and what information is shared, entails rather selective decisions that consider the potential effects of such (non-)disclosure on family members. This aligns with previous research showing that individuals seek the "right time" to discuss the subject with family members (Derbez, 2018; Forrest et al., 2003; Gaff et al., 2007; Wilson et al., 2004). Our review also found that conversations about an IGC, particularly between parents and children, typically unfold as a process and not a one-time event. This has been reported in previous reviews (Gaff et al., 2007; Wilson et al., 2004) and aligns with findings suggesting that patients prefer to share genetic information in a stepwise manner (van den Heuvel et al., 2019).

Our review also shows that individuals discuss various aspects of the IGC with their family members. While previous reviews have predominantly concentrated on the sharing of genetic risks (Ahsan et al., 2023; Gaff et al., 2007; Rowland & Metcalfe, 2013; Seymour et al., 2010; Srinivasan et al., 2020; Wilson et al., 2004; Wiseman et al., 2010), our review adds to this knowledge by highlighting that families also engage in discussions about health and reproductive implications, the genetic counseling process, impacts on children, and challenges in reaching out to specific relatives. Additionally, our review demonstrates that women often assume a pivotal role in the gathering and dissemination of information. In particular, some studies identified mothers and grandmothers are central in steering the family communication process, aligning with previous research (Ashida et al., 2011; Koehly et al., 2003, 2009).

A sense of responsibility and moral obligation toward family members, along with fear of discrimination and stigma, were identified as one of the main facilitators and barriers to family communication, respectively. These findings are generally consistent with existing reviews on family communication, which primarily focus on non-LOND families (Gaff et al., 2007; Seymour et al., 2010; Srinivasan et al., 2020; Wiseman et al., 2010). However, our review also identified that concerns about stigmatization and discrimination are significant barriers to discussing the condition with family members (Klitzman & Sweeney, 2011; Klitzman et al., 2007; Mendes et al., 2017; Oliveira et al., 2020; Wain et al., 2009), which has been described only minimally in previous reviews (Srinivasan et al., 2020; Wiseman et al., 2010). In their proposed framework on family information management, Abad et al. (2024) identify stigma and discrimination as contextual influences on that process. However, their review does not specifically address the aspect of avoiding discussions about the condition with family members. Additionally, and perhaps surprisingly, we found that only one study identified the absence of a cure for the IGC as a reason for not discussing it within the family (Pierron et al., 2021). In contrast, previous studies have suggested that the availability of risk-reducing interventions may foster greater openness in family discussions of genetic information (Dheensa et al., 2016; Forrest et al., 2003).

Our review also suggests that family communication spans a continuum ranging from closedness to openness, which is influenced by individual, interpersonal, and contextual factors. This process extends beyond the transmission of single biological facts, involving an assessment of the subjective experiences and the potential impact that the information may have on both the recipient and the transmitter. This communication is enmeshed in a familial context shaped by the family's shared experience of living with or being at risk of an inherited condition (Abad et al., 2024; Mendes et al., 2018; Rolland & Williams, 2005). Moreover, consistent with the family systems literature (Pantaleao et al., 2020; Young et al., 2019), our findings suggest that communicating about an IGC is influenced by family dynamics, including intergenerational patterns of organization (e.g., parents typically serving as gatekeepers of information, and women as gatherers and disseminators) and relationships (e.g., perceived closeness or emotional bonding, as well as conflict), along with transitions throughout the family life cycle.

Additionally, the dissemination of genetic information in the family typically begins with first-degree relatives and then moves into the extended family (Keenan et al., 2005). These communication patterns and preferences are generally consistent with research conducted in families facing other types of IGCs (Ashida et al., 2009; Koehly et al., 2009). This research

highlights that roles and norms are integral components of family scripts passed down through generations as part of the family narrative. These scripts shape how family members communicate about genetic risk, illness, and healthcare.

Overall, our findings highlight the intricacies of how families disseminate genetic information. This process may reflect recommendations received from genetic healthcare specialists to patients regarding the cascading of information within the family. Additionally, it may indicate that individuals generally feel closer to their "nuclear" family members (i.e., first-degree relatives) than to extended family. Furthermore, the flow of communication within families may also mirror lay constructions of family and kinship ties, which shape family members' perceptions of genetic risk (Keenan et al., 2005). These understandings are culturally sensitive and can vary widely even within groups that share similar cultural norms, and perceptions of "who is the family" (Atkinson et al., 2013). This will have profound implications for discussions about genetic risk and the identification of at-risk relatives in genetic counseling. As clinical trials for several LONDs advance and healthcare places greater emphasis on informing patients' relatives (Mendes & Newson, 2024; Nääs et al., 2024), particularly when effective treatments and prevention are available, this review underscores the importance of situating information-sharing within the relational dynamics of families, including gender, family structure, organization, and developmental transitions.

Finally, the majority of the included studies addressed exclusively families with HD. It was the first LOND for which predictive genetic testing became available (Brouwer-Dudokdewit et al., 2002). This development, along with the downstream effects of the availability of genetic testing, has likely facilitated subsequent studies on communication patterns and other social and behavioral research in families affected by or at risk for the disease. This trend may account for the prominence of studies involving families with HD.

This review has several limitations. Firstly, it is restricted to peer-reviewed publications in English, Spanish, Portuguese, or French, excluding any material not yet published, or published in other languages. Secondly, the included studies report the experience of families enrolled in research studies. Thus, it may not address the experiences of those who do not wish to participate in research. Third, there is considerable heterogeneity in the included studies on the terms used and the level of detail provided, e.g., regarding the sociodemographic characteristics of participants or their genetic risk status. It was therefore difficult to categorize information for both the descriptive analysis of the study characteristics and the content analysis of textual data.

The studies included in this review were all retrospective, primarily utilized a qualitative design, and predominantly focused on HD families. Future studies should be extended to other LOND to explore potential differences among conditions. This should include exploring how different clinical features and disease severity influence decision-making and communication patterns within families, as well as on disease and risk management, e.g., genetic testing uptake, reproduction decisions, or involvement in clinical trials for new drugs. Additionally, as new therapeutic options are expected to expand the clinical management of these conditions and clinical trials for testing new drugs continue to progress (Andrew & Fox, 2023), further research could explore how perceptions on the improving "manageability" of specific LONDs impact family communication. Future research should also explore family communication as a longitudinal process, employing study designs that capture ongoing interactions and incorporate the perspectives of multiple family members involved in the communication network. Finally, research stemming from different countries with more diverse population groups should be encouraged, as how individuals make decisions about communication with family members certainly differs among countries, regions, and populations.

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ORCID

Sofia Fontoura Dias http://orcid.org/0000-0002-2938-7486
Maria Barbosa http://orcid.org/0009-0008-3590-3847
Filipa Júlio http://orcid.org/0000-0001-6075-6887
Angus Clarke http://orcid.org/0000-0002-1200-9286
Alison Metcalfe http://orcid.org/0000-0002-6466-918X
Jorge Sequeiros http://orcid.org/0000-0002-9846-1037
Milena Paneque http://orcid.org/0000-0002-6535-4315
Liliana Sousa http://orcid.org/0000-0003-3491-7119
Álvaro Mendes http://orcid.org/0000-0002-8766-7646

Author contributions

SFD was involved in study design, data extraction, analysis, and manuscript writing and editing. ÁM contributed to manuscript writing and editing. ÁM and MB contributed to data extraction, and MB, FJ, LS, and ÁM contributed to the study design and data analysis. All authors critically edited the manuscript.

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