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REVIEW ARTICLE

Exploring stigma in Huntington's disease: A scoping review of methods and conceptualizations for understanding experiences of gene expansion carriers and at-risk individuals

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Abstract

Many individuals affected by the hereditary neurological condition Huntington's disease (HD) have reported experiences of stigmatization, yet the extant literature is currently theoretically and methodologically underdeveloped. Therefore, this scoping review aimed to examine the methodological approaches and theoretical conceptualizations of stigma used and to identify areas of underrepresentation given the emerging evidence base. A systematic search of five databases and hand-searches of included papers resulted in 3273 articles. Thirty-two met predefined inclusion criteria, which included primary research reporting experiences of stigma by people with HD or who have an inherited genetic risk. Selected articles represented qualitative (59% of included papers), quantitative (22%), and mixed-methods (19%) designs. Almost half did not provide theoretical definitions of stigma. Theories describing societal unacceptance based upon a discrediting attribute of an individual by the sociologist Erving Goffman, and of discrimination solely based upon real/perceived differences from the typical genome, were most prominent among theories referenced, conceptualizing stigma and genetic discrimination respectively. Findings suggested a limited theoretical grounding of stigma in primary research exploring HD, with a lack of discussion around conceptualizations of stigma applied to people affected by HD. Future research should apply clear definitions to differentiate constructs contributing to different forms of stigma and could explore whether experiences of stigma and support needs might differ across groups affected by HD. Implications for theoretical development and multilevel interventions are also discussed.

KEYWORDS
conceptualization, Huntington's disease, scoping review, stigma

1 | INTRODUCTION

Huntington's disease (HD) is a hereditary neurodegenerative disease, with an estimated global pooled prevalence of 4.88 cases per 100,000 people (Medina et al., 2022). HD affects cognition, movement, emotions, and behavior. Diagnosis follows the onset of visible motor symptoms ("manifest HD") which typically occurs between 30 and 50 years (Walker, 2007). No cures or disease-modifying

therapies are presently available, and the mean duration from disease onset to death is around 20 years (Roos, 2010). Individuals with nonspecific motor symptoms and clear cognitive changes may be referred to as "prodromal HD" (Medina et al., 2022), while those who have tested positive for the HD gene without symptoms are referred to as presymptomatic or premanifest. Individuals with a parent with HD who have not undergone genetic testing are referred to as "at risk" (Medina et al., 2022). The term "people affected by HD" will be used throughout this review to refer to HD gene expansion carriers, individuals clinically diagnosed with HD, and those at genetic risk with a parent diagnosed with HD (i.e., including all those who carry the gene expansion, plus those at risk). However, as will be evident throughout the rest of this review, stigma does affect individuals within these groupings differently and where data allow, we will indicate this.

People with manifest HD experience difficulties with motor function, balance and coordination, including sudden involuntary movements ("chorea"), and difficulties with speaking and swallowing. These challenges typically reduce independence, can be life-threatening, and often affect individuals emotionally. Moreover, they engender additional social problems. For example, such visible difficulties may appear unusual to others and may be misunderstood as intoxication, attracting unwanted attention and stigmatization (Etchegary, 2007). Cognitive abilities (e.g., concentration, problem-solving and memory) are also impacted by HD and, alongside emotional difficulties including anxiety, low mood, irritability (e.g., Novak & Tabrizi, 2011; Roos, 2010), and perseveration can lead to behaviors that others may judge negatively. Such challenges can make social participation harder for people with HD, affecting communication, emotional regulation and accessibility (which may occur before motor symptoms begin, e.g., Gibson et al., 2022).

What is known about this topic

Symptoms of Huntington's disease (HD) such as an unsteady gait or slurred speech may engender stigma, as others may struggle to understand or accept these differences. However, while many people affected by HD have reported experiences of stigmatization, the extant literature has reported disparate and vague conceptualizations of stigma within empirical research.

What this paper adds to this topic

This paper provides a comprehensive scoping review of research on stigma experienced by individuals affected by HD. It highlights key gaps, including the lack of conceptual clarity and theoretical frameworks around stigma in many studies. Despite a growing interest in the topic since 2008, stigma has rarely been a central research focus and most findings have been incidental, limiting meaningful exploration. This review recommends more rigorous, longitudinal studies, and the use of validated stigma measures. It also highlights the importance of examining stigma at societal and structural levels, advocating for a shift away from individual-focused approaches. This review emphasizes the need for a more cohesive theoretical framework and targeted interventions to address stigma's impact on people with HD.

1.1 | Stigma and HD

Perceptions and lived experiences of stigma may increase as symptoms become more visible, as others may struggle to understand or accept these differences (Sherman et al., 2020). In 1963, the influential sociologist and stigma theorist Erving Goffman defined stigma as a "deeply discrediting" (p. 3) attribute resulting in societal unacceptance. Building upon Goffman's theory, Crocker et al. (1998) argued "stigmatized individuals possess (or are believed to possess) some attribute, or characteristic, that conveys a social identity that is devalued in a particular social context" (p. 505), suggesting stigma is context-dependent. Indeed, social norms can become a context for stigmatization. For example, negative portrayals and attitudes toward disability and its burden are perpetuated across numerous cultures (e.g., in India, Banga & Ghosh, 2017; USA, Green, 2003; and Hong Kong, Ngan et al., 2020). Consequently, families of children with a disability have reported stigma, marginalization and associated feelings of shame and isolation, increasing the difficulty of support-seeking (e.g., Lam & Mackenzie, 2002; Ngo et al., 2012).

Adding further nuance to "vaguely defined and individually focused" conceptualizations of stigma, Link and Phelan (2001) acknowledged the identification of human differences as a social process, moving away from locating stigma as an attribute within the person (p. 363). They discussed five components necessary for the presence of stigma: "labeling" differences considered socially relevant; associating these differences with negative attributes; separation of "us" from "them"; resulting discrimination and loss of status; and the dependence of stigma on the stigmatizing group holding power. For instance, individuals experiencing sudden involuntary movements may be labeled and stereotyped by others in undesirable ways, thereby separating them from others without such characteristics. Consequently, Link and Phelan (2001) advised that this can result in the person being placed lower in social hierarchies, representing a loss of status and increased potential for discrimination (e.g., rejection for a job based upon the perceived difference of the person with motor symptoms). In short, stigma is therefore conceptualized as existing when people distinguish human differences according to perceived undesirable characteristics, diminishing the individual's social status and access to social, economic or political power.

Further consequences of stigma have been identified, whereby stigma has been noted as a global barrier for health-seeking behavior

or treatment adherence for various conditions including HIV/AIDS (Mahajan et al., 2008), epilepsy (Dilorio et al., 2003), lung cancer (Scott et al., 2015), and mental health (Corrigan, 2004). It affects health outcomes and quality of life by undermining social relationships, access to healthcare, housing, and employment, while increasing stress and perpetuating social injustice (Hatzenbuehler et al., 2013; Stangl et al., 2019).

People with neurological conditions experience stigma particularly when symptoms cannot be concealed; in the context of HD, symptoms such as an unsteady gait or slurred speech may engender stigma (Rao et al., 2009). Medical sociologist Graham Scambler (1998) discussed stigma within the context of disease, introducing a categorization of stigma distinguishing between enacted (regarding actual social rejection/discrimination) and felt stigma (reflecting one's fear of future discrimination). While Link and Phelan's (2001) conceptualization concerns differences perceived by others, Scambler posited that felt stigma is typically more disruptive than enacted stigma, as people tend to conceal their illness and symptoms thus reducing opportunities for others to stigmatize noticeable difficulties. Yet, for people with manifest HD, visible symptoms are often unavoidable. Felt stigma may also lead to a person internalizing stigma by identifying with negative stereotypes, and experiencing diminished self-esteem and self-efficacy ("self-stigma") for people with "mental illness" labels (Watson et al., 2007).

Given that predictive genetic testing is available for those with a family history, stigma around those who carry the HD gene without visible symptoms is also possible. Developments in genetic testing raised concerns regarding the potential stigmatization of families and individuals affected by genetic illness, as individual responsibility for health became embedded within social and historical contexts, with connotations of blame and culpability for those affected (Harper, 1992; Savulescu & Kerin, 1999). Alarmingly, Elger and Harding (2003) surveyed 599 students and reported that 73.2% of law students and 39.4% of medical students sampled in Switzerland agreed that society should do everything possible to reduce the frequency of HD, including social pressure to recommend genetic testing and sterilization. Therefore, attitudes which could be interpreted as eugenic appear to have endured, which may understandably prevent individuals from pursuing genetic counseling. Indeed, concerns about the misuse of genetic information persist, with documented cases of genetic discrimination against people with HD (e.g., Bombard et al., 2012). Genetic discrimination is defined as adverse treatment against an individual or their family members based on real or perceived genetic differences (Billings et al., 1992; Gostin, 1991). Therefore, families affected by HD may face societal intolerance and blame, even before symptoms appear. Thus, individuals or families affected by HD might limit their social interactions or hide their condition due to stigma, missing out on support that genetic counseling might otherwise provide. Therefore, stigma, in all its forms, can lead to social inequalities for all people affected by HD, negatively impacting social participation, quality of life, and access to opportunities.

Substantial research has explored experiences of genetic testing among participants affected by HD. This body of research includes reasons for and against genetic testing (e.g., Binedell et al., 1998; Decruyenaere et al., 1995; Paneque et al., 2019), communication regarding genetic risk (e.g., Rowland & Metcalfe, 2013), and the impact of testing such as within employment and insurance sectors (e.g., Oster et al., 2008; van der Zwaan et al., 2022). However, fewer studies have explicitly aimed to explore personal experiences of stigma from people at risk of, or with HD. Link and Phelan (2001) highlighted how theories of stigma should be informed by the lived experience of the stigmatized to avoid misunderstanding their experiences and perpetuating unsubstantiated assumptions.

1.2 | Rationale for current review

Initial scoping of the area of interest demonstrated disparate and vague conceptualizations of stigma within empirical research, as some explored singular constructs of stigma (e.g., genetic discrimination) while some did not explicitly define stigma using theoretical definitions. This was a pervasive issue across the literature identified, and this will be discussed further as part of this scoping review. Moreover, studies included qualitative, quantitative and mixed methodological approaches, with some studies only briefly reporting experiences related to stigma in their results. Accordingly, it was considered useful to explore this broad body of research. Given these disparate methodologies and conceptualizations of stigma, a scoping approach was considered useful to explore this broad body of research. Therefore, this scoping review aimed to explore the breadth of research discussing experiences of stigma and related concepts (such as genetic discrimination) among people affected by HD, to offer directions for meaningful future research.

Scoping reviews aim to map the existing literature surrounding a topic of interest, exploring the volume and characteristics of primary research (Arksey & O'Malley, 2005; Pham et al., 2014). A common reason for conducting a scoping review is to explore the breadth of existing literature, with a specific focus on, for example, the methodologies used and key reported concepts, to identify knowledge gaps and guide directions for future research (Peters et al., 2020). The heterogeneity of methodologies and underrepresentation of theory-driven constructs related to stigma in the currently existing literature precludes a systematic review at this stage. Therefore, a scoping review was considered necessary to provide a critical exploration of the concept to stimulate further research to be more theoretically informed.

Therefore, the present scoping review investigated:

1. What are the key concepts and definitions of stigma (including the construct of genetic discrimination) reported in primary research investigating people affected by HD?
2. What methodological approaches have been utilized to explore stigma?
3. What are the subsequent gaps or areas of underrepresentation (relating to populations, constructs and/or evidence)?

2 | METHODS

Arksey and O'Malley (2005) described a five-stage framework for conducting scoping reviews (subsequently further developed by Levac et al., 2010): (1) identifying research questions; (2) identifying relevant studies; (3) study selection; (4) charting the data; (5) collating, summarizing and reporting results. Both inform the Preferred Reporting Items for Systematic Reviews extension for Scoping Reviews (PRISMA-ScR; Tricco et al., 2018), which includes a reporting checklist (included in the Appendix S1) that guided the present review. An assessment of methodological limitations or risk of bias in individual papers is usually not performed within scoping reviews (Pham et al., 2014). The protocol was developed to reflect the stages and reporting items described above.

2.1 | Identifying the research question

A preliminary literature search on Google Scholar and APA PsycINFO was conducted to determine whether a similar scoping review had been published and to clarify the breadth of published study aims, methods, and findings. The question, "What are the key concepts and definitions of stigma reported in HD research?" guided the search strategy, while methodological approaches and areas of underrepresentation within the literature were identified post hoc.

2.2 | Identifying relevant studies

A specialist subject librarian was consulted for support with refining the search strategy, including selection of keywords, Medical Subject Headings (MeSH), and database selection. Preliminary searches and consultations with the librarian produced relevant search terms (Table 1) and eligibility criteria (Table 2). The decision was made to include all papers reporting on individuals affected by HD, from at-risk to symptomatic. Only publications from 1993 onwards were included as predictive testing became available from this date (MacDonald et al., 1993).

A systematic literature search was performed in July 2023, using the same search terms across the following databases: APA PsycINFO, Medline, Cumulative Index to Nursing and Allied Health Literature (CINAHL), Scopus and Web of Science Core Collection. Terms relating to stigma and HD were exploded using MeSH, then search terms were inputted. The full strategy for Medline is depicted in the File S1. Database filters were not applied before screening, reducing the risk of excluding eligible records.

2.3 | Study selection

A total of 3273 articles were obtained from five databases (Figure 1). Reference lists of included papers were hand-searched, which did not identify further eligible papers. Citations were imported into Endnote for duplicate removal ($n = 1180$), before importing remaining articles to Rayyan (a bibliographic software program) where titles and abstracts were screened using the inclusion and exclusion criteria. After screening abstracts and titles, full texts for 151 papers were accessed for further screening, resulting in 32 papers selected for inclusion. Five articles that did not meet inclusion criteria and five articles that did were randomly selected and reviewed against eligibility criteria by a psychologist trained in research methods. There were no discrepancies in study selection.

It was noted that some papers analyzed data from the same samples. Klitzman (2010), Klitzman and Sweeney (2011) and Klitzman et al. (2007) analyzed data from the same sample of 21 people affected by HD; Wauters and Van Hoyweghen (2018, 2021) included data from an overlapping sample; Erwin et al. (2010) and Williams (2010) also analyzed data from the same sample; and Bombard et al. (2009, 2011, 2012) analyzed quantitative data from the same sample ($n = 233$). All of these papers were included as, while they drew on the same or similar samples, the papers had different areas of focus, thus presenting meaningfully different data from the samples and not simply

TABLE 1 Search terms applied in systematic search strategy.

Population	Stigma
huntington*	stigma* or "health-related stigma" or discriminat* or "genetic risk*" or prejudic* or injustice* or devalu* or dehumani?ed. or stereotyp* or shame* or shaming or blame or blaming or taboo* or inequalit*

Note: " = search phrase; * = truncation, that is, different word endings; ? = wildcard symbol, that is, different spellings/characters. Search terms from the columns were combined for the final search using the Boolean operator "AND."

TABLE 2 Inclusion and exclusion criteria.

Inclusion	Exclusion
Individuals with the HD gene expansion or at-risk Published primary research articles describing definitions, experiences or outcomes related to stigma, or gray literature if equivalent findings are not published in peer-reviewed journal English language From 1993 Any country Qualitative, quantitative, or mixed methodology	Only reports perspectives of healthcare providers, caregivers or family members not at risk of HD (i.e., no data from the population of interest, or data from the population of interest not reported separately from others) Articles without an aim to explore definitions, experiences or outcomes related to stigma, or without describing experiences or outcomes related to stigma in the results from the population of interest Protocols Abstracts, poster, or conference presentations without published full-text Editorials, letters, or reviews that do not present new data

Abbreviation: HD, Huntington's disease.

replicating the results from previous analyses on data from the same sample. Where this was not the case (e.g., Etchegary, 2005), then the paper was excluded.

2.4 | Charting the data

Relevant data were extracted from the selected papers and collated using Microsoft Excel (see Appendix S1 for study characteristics). All data relating to stigma in the context of HD were sought and collated from the papers, with a broad scope appropriate to the scoping review approach and the research questions. Consequently, data could be taken from any section of the paper. Regarding coding, this was largely carried out by the first author and most of the data were extracted and summarized descriptively given the research questions. The specific research questions were considered to direct the extraction of the data, and so the analysis could be considered deductive. The other authors screened around 20% of the papers selected for inclusion to ensure that the first author had summarized and extracted appropriately. There were no significant discrepancies between the first author and the other authors during this process; where minor discrepancies occurred, these were resolved within the team and mostly related to the level of detail needed within the Tables.

3 | RESULTS

Thirty-two articles were included; these were published between 1993 and 2023, representing 13 countries and 3781[1] participants. Eleven papers reported samples exclusively from the United States of America (USA), seven from Canada, three from Australia, two from Belgium, two combining participants from the USA, Australia, and Canada, one from England and Wales, one from Norway, one from Spain, one from Portugal, one from Colombia, one combined sample from Germany, Austria, and the USA, and one from China. Several sets of authors contributed multiple papers (e.g., Bombard et al., Carlozzi et al., Klitzman et al., and Wauters & Van Hoyweghen – see Methods above for details).

Following the framework developed by Arksey and O'Malley (2005), a narrative overview of the selected records is presented. Reporting focused on the three study aims of identifying the key concepts and definitions of stigma reported in HD research, exploring the methodological approaches used to explore stigma in HD populations, and gaps and/or areas of underrepresentation in this area. These will be addressed sequentially. Summaries of study characteristics and key findings are presented in Tables 3–5; please also refer to the expanded versions in the File S2 for more detail.

[1]This figure refers to the total number of participants reported across the papers. It does not refer to the total number of unique participants, as this is not possible to report given that some samples were clearly overlapping but the unique number in each was not clear.

3.1 | Key concepts and definitions of stigma in HD research

Overall, 10 papers explicitly provided theoretically grounded conceptualizations of stigma in the introduction to orientate the research and/or to direct their analysis. Six described theories by Goffman (1963), three referred to Link and Phelan (2001), two cited Crocker et al. (1998), two cited Gostin (1991), two referred to Scambler (1998), and one to Geller et al. (1996). Seven (50%) qualitative papers referenced theoretical definitions of stigma, as did one (14%) quantitative study, and two (33%) mixed-methods papers. Some papers referred to multiple theories of stigma.

Eleven papers provided theoretical definitions of genetic discrimination – seven (37%) qualitative papers, four (43%) quantitative papers, and one (17%) mixed-methods paper. Ten applied Billings et al.'s (1992) definition, and one applied Rothstein and Anderlik's (2001).

Six described theoretical underpinnings of both stigma and genetic discrimination, acknowledging that these concepts are intertwined. Fourteen papers (44%) did not state a theoretical definition or conceptualization of stigma or related concepts (in these cases, relevant experiences were identified in the results, hence the inclusion in this review).

Four qualitative papers, one mixed-methods and one quantitative study explicitly described theoretical definitions of stigma in relation to living with HD, again referencing Goffman (1963). Two papers referred to Goffman in their introductions and discussions, one paper only in their introduction, thus theoretically orientating readers but without applying theoretical concepts of stigma when interpreting results. Three papers applied Goffman's theories while analyzing results, expressly anchoring findings to conceptualizations of stigma.

Several less commonly cited conceptualizations of stigma were referenced. Three papers referred to Link and Phelan's (2001) aforementioned conceptualization of perceived undesirable characteristics, whereby people perceived as different are placed in an out-group, resulting in a loss of social, economic, or political power. Boileau et al. (2020) referred to felt and enacted stigma in their introduction, although they did not cite Scambler's (1998) underpinning theory; Wauters and Van Hoyweghen (2021) also introduced interpretations of felt and enacted stigma in their deductive analysis to develop meanings within the results. Etchegary (2007) expressly referenced these constructs, including using them to ground the qualitative analysis within the thematic structure.

Eighteen papers explored experiences of genetic discrimination, mostly among people at risk of HD. Ten papers used the theoretical definition by Billings et al. (1992) to operationalize this construct, referring to differential treatment of asymptomatic people according to genetic differences rather than physical/observable features. Most papers referring to Billings et al. (1992) only included individuals who were asymptomatic but at risk for HD. However, Wauters and Van Hoyweghen (2018) included two symptomatic participants (14.3% of the sample), suggesting the conflation of models of discrimination, as some participants may have reflected upon discrimination related to visible symptoms or disease progress rather than genetic traits.

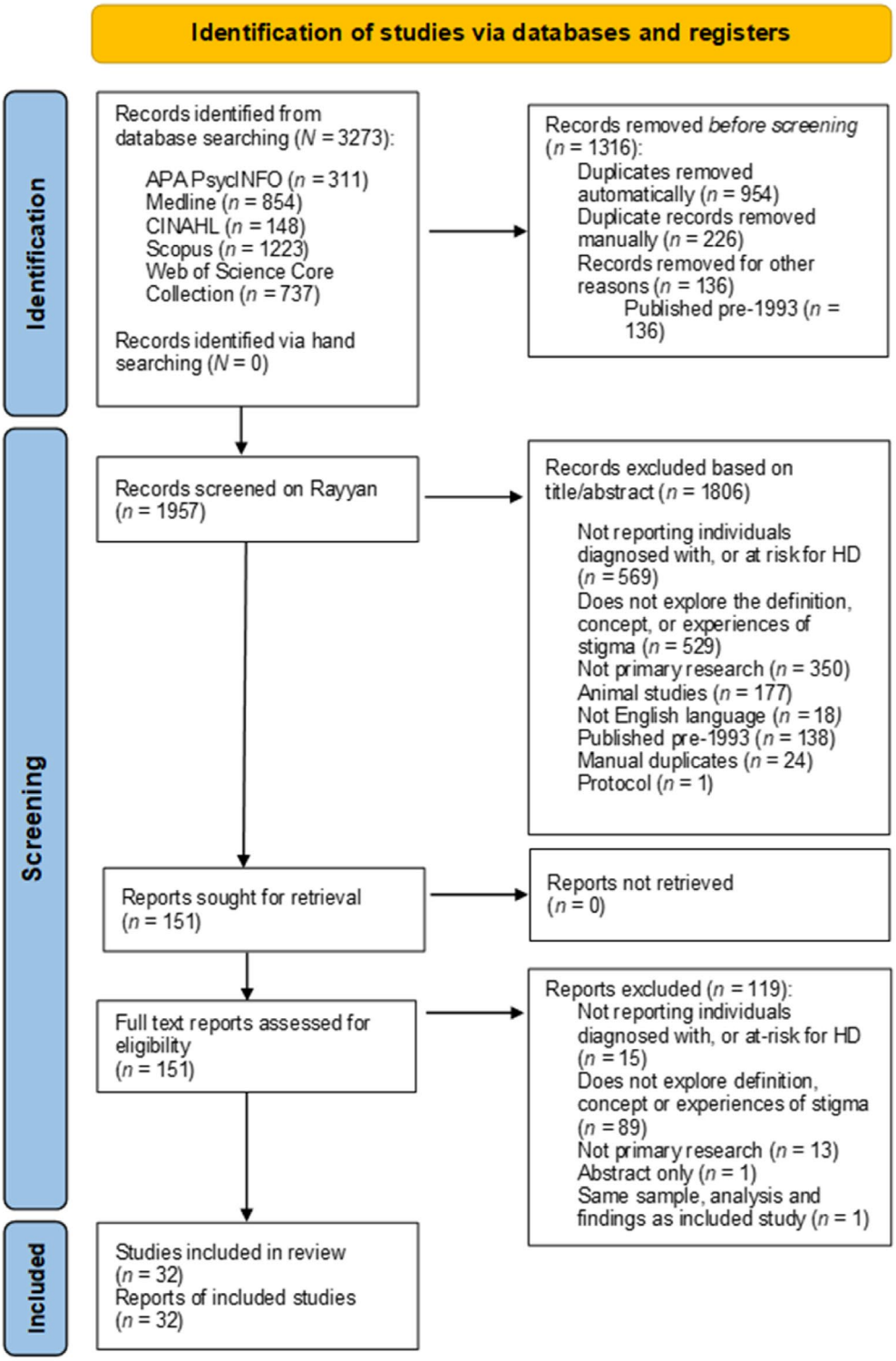

FIGURE 1 PRISMA flowchart of study selection (adapted from Page et al., 2021).

Other conceptualizations of genetic discrimination were less frequently cited, with one paper citing Gostin's (1991) definition, and one following Rothstein and Anderlik (2001) in describing genetic discrimination as adverse treatment based solely on the family history or genotype of people without symptoms of a disease. While the term "genetic discrimination" was used frequently in Klitzman (2010), the author defined discrimination using the Oxford English Dictionary (1993): "…an act of distinguishing…discriminating against people on grounds of race, color, sex, social status…; an unjust or prejudicial distinction," broadening their focus of study significantly from genetic discrimination as understood by other definitions noted above.

In sum, a range of conceptualizations of both stigma and genetic discrimination were applied across the retrieved papers. Some

TABLE 3 Summary of qualitative study characteristics and key findings.

Author (year)	Country of origin	Sample (participants meeting criteria for review)	Aims	Methodology	Type(s) of stigma explored, including definition (if given)	Findings related to stigma
Bombard et al. (2007)	Canada	37 asymptomatic individuals with positive genetic status for HD 62.2% female	Explore how people with HD gene expansion use strategies to manage risks and experiences of GD	Semi-structured interviews (telephone and face-to-face) Grounded theory analysis	GD defined as differential treatment of asymptomatic people according to genetic differences rather than physical features (Billings et al., 1992)	Four coping strategies identified: *"Keeping low"*: keeping genetic testing/family history of HD private *"Minimizing GD"*: disregarding GD encounters, avoiding confrontation *"Pre-empting GD"*: action to evade or protect self from GD *"Confronting GD"*: directly resisting or challenging experiences
Bombard et al. (2008)	Canada	45 genetically tested 10 untested at-risk individuals 67% female	Describe concerns and experiences of GD among people at risk for HD	Semi-structured interviews (telephone and face-to-face) Grounded theory analysis	GD defined as differential treatment of asymptomatic people according to genetic differences rather than physical features (Billings et al., 1992)	Concerns about and experiences of GD in family, insurance, employment, social, healthcare, and government domains Feeling "tainted" by family history of HD in social and employment contexts Awareness of GD precipitated by adverse consequences of genetic difference (e.g., family members' experiences)
Etchegary (2007)	Canada	20 individuals at-risk of HD 4 family members Age in years M (SD): 46 (11.3) 75% female	Explore participants' perceptions of stigma related to HD	Semi-structured interviews (telephone and face-to-face) Interpretative phenomenological analysis	Enacted stigma: actual social rejection or discrimination "Felt stigma": feared future discrimination (Scambler, 1998) GD: discrimination based on real or perceived differences from norm (Billings et al., 1992)	Four themes presented: *"Sympathy, not stigma"*: felt stigma; others being sympathetic or ignorant about HD rather than overtly stigmatizing *"Genetic discrimination"*: difficulties obtaining life or health insurance *"Enacted stigma"*: impact of avoiding public negative responses from others on behavior and social relationships *"Dismissing stigma-ignorant others"*: not internalizing others' stigmatizing attitudes, seen as reflecting ignorance of HD

(Continues)

TABLE 3 (Continued)

Author (year)	Country of origin	Sample (participants meeting criteria for review)	Aims	Methodology	Type(s) of stigma explored, including definition (if given)	Findings related to stigma
Hamana et al. (2021)	England and Wales	25 people with prodromal to late-stage HD (32% female) 25 family caregivers (76% female)	Explore how living with HD impacts physical activity across HD stages; identify theoretical framework to develop physical activity interventions	Focus groups on participation and motivation within physical activity Framework analysis	Stigma not described Acknowledgment that stigma affects people with HD with a cost to quality of life	Stigma of HD: public and family relationships, other people's lack of understanding or knowledge of HD and its consequences for physical activity, and stigma/breakdown/difficulties in family relationships due to HD Feeling stigmatized in public when participating in physical activity, attributed to HD symptoms (e.g., unstable gait and chorea)
Kjoelaas et al. (2022)	Norway	36 offspring with HD caregiver (age range 13–65 years; M: 36.6; 72.2% female) 14 caregivers with children with an HD partner (age range 42–67 years; M: 54.9; 78.6% female)	Explore caregiver and offspring perspectives on HD throughout childhood	Individual semi-structured interviews Thematic analysis	Not explicitly stated; stigma discussed as theme in results	*"To share or not to share?"*: dilemmas regarding sharing HD status. Subtheme *"Help or stigma?"*: offspring's reflections on talking about HD; often encountered negative/stigmatizing responses Not receiving support and understanding causes feelings of rejection. Offspring feared HD stigma would hinder obtaining insurance or workplace promotions
Klitzman (2010)	USA	21 participants: 7 untested 10 gene-positive (6 symptomatic) 4 gene-negative 42.86% female 85.7% White; 9.5% Black; 4.8% Hispanic	Explore how HD impacts experiences of discrimination, including concerns, impact on health decisions and behaviors	Individual semi-structured interviews Grounded theory	GD, defined as "…an act of distinguishing… discriminating against people on grounds of race, color, sex, social status…; an unjust or prejudicial distinction" (Oxford English Dictionary, 1993)	*"Difficulties with Discrimination"*: explicit and implicit; legal protection may be limited *"Implications"*: GD concerns can impede genetic testing, treatment, career and life decisions; related stress may exacerbate illness Evidencing GD can be difficult as nongenetic factors may influence others' treatment (e.g., in employment) Psychiatric symptoms can produce stigma and discrimination (e.g., mood and behavior changes linked to HD)

TABLE 3 (Continued)

Author (year)	Country of origin	Sample (participants meeting criteria for review)	Aims	Methodology	Type(s) of stigma explored, including definition (if given)	Findings related to stigma
Klitzman et al. (2007)	USA	21 participants: 9 untested 8 gene-positive 4 gene-negative 42.86% female 85.7% White 9.5% Black 4.8% Latino	Explore experiences of being at-risk for HD, genetic testing, learning one's genetic status; explore disclosures of HD risk within families	Individual semi-structured interviews Grounded theory	Stigma associated with disclosing genetic status, defined per Goffman (1963); people with stigmatized conditions try to manage "tainted" (p. 3) features of their identity and "pass" (p. 80) as "untainted" (p. 3)	Concealing symptoms viewed as evidence for stigmatization; viewing oneself as "abnormal" may affect disclosures, sharing partial, indirect, or no information Mental health symptoms stigmatized; HD a *"big secret in the family"* Recent attitudes toward HD seen as changing—older generations may have been more secretive, engendering less understanding and more stigma
Klitzman and Sweeney (2011)	USA	21 participants: 9 untested 8 gene-positive 4 gene-negative 42.86% female 85.7% White; 9.5% Black; 4.8% Latino	Explore reported dilemmas within dating and relationships related to disclosing genetic risk status	Individual semi-structured interviews Grounded theory	Stigma associated with disclosing genetic status, defined per Goffman (1963); people with stigmatized conditions try to manage "tainted" (p. 3) features of their identity and "pass" (p. 80) as "untainted" (p. 3)	Individuals at genetic risk do not try to "pass," but try to avoid stigma and rejection to establish a relationship. Fear of rejection discouraged disclosures, with ethical conflict: balancing privacy against sense of responsibility to disclose Reported feelings of "embarrassment" (p. 108) when disclosing family history to partners
Leung and Leung (2002)	China	Five Chinese families from Hong Kong; recruitment strategy not reported	Review psychosocial aspects of Chinese families with HD	Interviews and analysis of case records Results reported narratively	"Social stigma," not further described beyond noting symptoms of HD which "scare people off" (p. 308)	One individual dismissed from work due to excessive movements; wife avoided taking him out, "fearing stigmatization" One stayed single due to prognosis; advised to avoid women lest his movements be mistaken as unfriendly or indecent, suggesting social isolation and stigma
Oliveira et al. (2020)	Portugal	4 participants from HD families: Two tested negative Two tested positive Age in years M (SD): 31.75 (4.11) 100% female Sample obtained from Oliveira et al. (2017)	To explore how families affected by HD manage information about HD from a transgenerational perspective	Semi-structured interviews (telephone, Skype or face-to-face) Thematic analysis	Stigma described as reported fears of stigma and discrimination leading to secrecy and shame. Participants' response to stigma and discrimination discussed as "resistance to being treated differently" (p. 1215)	Three themes: "searching for a diagnosis"; "making sense of HD in the family"; "managing HD in the family" Reports of transgenerational stigma, taboo, fear of others' responses, and shame; secrecy-based strategies used to conceal HD-related symptoms to avoid stigma Shame and stigma suggested as attached to experiences of HD in families

(Continues)

TABLE 3 (Continued)

Author (year)	Country of origin	Sample (participants meeting criteria for review)	Aims	Methodology	Type(s) of stigma explored, including definition (if given)	Findings related to stigma
Penziner et al. (2008)	USA	15 individuals tested positive for HD gene expansion Age range: 22–62 years 73.33% women	Develop and test a qualitative approach to explore perceptions of GD in people testing positive for HD	Semi-structured interviews via telephone Content analysis, presented numerically to reveal patterns and gauge magnitude of concerns reported	Stigmatization defined as a person being treated differently in activities and socially due to their genetic status (Geller et al., 1996). GD defined as bias against a person or family due to real or perceived deleterious genetic status (Billings et al., 1992; Treloar et al., 2004)	GD and stigmatization concerns categorized into employment, insurance, and relationship contexts 90% of employed participants reported low concern regarding employment discrimination and had revealed test results to employers. 88.89% would not disclose if seeking new employment Instances of work discrimination shared 83% of insured participants did not disclose test results to insurers; 91% concerned about impact of test results on future insurability All participants perceived differential treatment following disclosure of test results with family members. 93% reported at least one negative impact on familial relationships
Quaid et al. (2008)	USA	55 people at-risk for HD 69.09% female Age in years M (SD): 42 (7.6) 87% White, 7% Black, 4% Hispanic, 2% Other PHAROS subset; did not differ from main sample	Explore the everyday experience of being at-risk for HD	Descriptive; open-ended unstructured interviews Naturalistic enquiry	Stigma not defined, but experiences related to stigma discussed in the results	"Living at risk" as overarching pattern; two foundational themes: concealment as an act of self-preservation, and preserving hope Differing experiences of disclosure: one person felt colleagues would protect her from others' stigmatization; another experienced discrimination in relationships, at work and a blocked promotion
Rivera-Navarro et al. (2015)	Spain	27 adult children of a parent with HD Age in years M (SD): 37.9 (6.7), range 24–53 55% female	Investigate factors influencing the decision to undertake genetic testing, among adult offspring of people with HD	Descriptive: qualitative sequential discourse from four focus groups	Stigma not defined, but experiences related to stigma discussed in the results	Four themes constructed: (1) Awareness of genetic testing; (2) In favor of genetic testing; (3) Against genetic testing; (4) Factors influencing decisions to take or not take the genetic test Stigma of HD in Spanish society discussed in all groups; heritability discourages testing, as others could infer that other family members may have HD

TABLE 3 (Continued)

Author (year)	Country of origin	Sample (participants meeting criteria for review)	Aims	Methodology	Type(s) of stigma explored, including definition (if given)	Findings related to stigma
Sherman et al. (2020)	USA	24 people with HD (8 early, 16 late) 15 with a parent with HD pretesting or prodromal for HD Age in years M (SD): 44.68 (12.06) 61.5% female 90% Caucasian, 5% African American, 2.5% Asian, 2.5% Other	Explore the impact of chorea on everyday functioning and health-related quality of life; identify patterns of perceptions and experiences of chorea	Focus groups comprising people with HD and people at-risk Thematic content analysis	Stigma not defined beyond considering impacts of chorea on social well-being: "chorea is often stigmatizing as they are commonly mistaken for drunkenness"	Global theme "Living with Chorea" described impacts of chorea on experiences of HD; "Stigma of chorea" as prominent subtheme At-risk participants feared potential stigma in future; those with manifest HD described experiencing stigma
Stuttgen et al. (2020)	USA	39 people affected by HD: 15 positive genetic test 21 negative genetic test 3 unknown test result Mean age in years: 59.6 51.28% female	Examine if and when individuals communicate HD risk, decide to undergo genetic testing, and share test results with others	Qualitative, semi-structured interviews, settings not reported Grounded theory	Stigma not described or defined; "discrimination" discussed in results	Participants selective about discussing their risk of HD, fearing discrimination or being viewed differently. Many did not inform colleagues or bosses, and typically did not disclose if they received a negative test result. Gene-positive people often waited until symptoms started to inform bosses
Varela et al. (2022)	Colombia	33 participants: 5 at-risk 5 reported as 'in anosognosia' 6 initial phases 14 intermediate phases 3 late phases 57.58% male Age in years M (SD): 42 (12)	Analyze how HD affects daily lives of individuals at-risk/diagnosed with HD; analyze the process of loss of sense of self	Qualitative, semi-structured interviews Grounded theory	Stigma described as being endured by people with HD, particularly when bodily changes are more noticeable (e.g., chorea), using Goffman's (1963) definition of stigma as a situation in which a disabled person does not receive full social acceptance	Core category identified as "Loss of sense of self until death." Five sub-categories: Context; Loss of sense of self; Relationship; and Daily life Discrimination experienced within relationships
Wauters and Van Hoyweghen (2018)	Belgium	9 tested positive for HD gene expansion 1 tested negative 4 indeterminates 78.57% female	Understand experiences and concerns regarding possible GD	Qualitative semi-structured interviews Grounded theory	GD defined as 'discrimination directed against an individual or family based on an apparent or perceived variation from the "normal" human genotype' (Billings et al., 1992; Natowicz et al. 1992)	Belgium legally prohibits the use of genetic information in insurance, but concerns remained about GD even among those aware of this legislation Participants concerned about GD and stigma for HD families, including indirect forms in work, relationships, and socially Some feared not being hired or promoted following a positive test result

(Continues)

TABLE 3 (Continued)

Author (year)	Country of origin	Sample (participants meeting criteria for review)	Aims	Methodology	Type(s) of stigma explored, including definition (if given)	Findings related to stigma
Wauters and Van Hoyweghen (2021)	Belgium	7 tested positive for HD gene expansion 1 tested negative 4 indeterminate Age in years M: 41, range: 28–60 83.33% female	Explore origins and sources of fears regarding GD, and how people cope	Qualitative semi-structured interviews Grounded theory	GD defined as "discrimination directed against an individual or family based on an apparent or perceived variation from the "normal" human genotype" (Billings et al., 1992; Natowicz et al. 1992). Stigma follows Goffman's (1963) categorization: a person is discredited when stigma is visible or known to others which taints them, linked to chorea. Discrimination and stigmatization noted as intertwined	Fears of GD suggested as resulting from experiences of stigmatization and discrimination against affected family members. Concerns about subtle and indirect forms of GD due to family history Some kept genetic status concealed to avoid stigma and GD
Williams et al. (2010)	USA, Australia, Canada	433 individuals at-risk for HD 71.2% female 61.8% from USA, 27.2% from Australia, 11.0% from Canada Age in years M (SD): 43.44 (10.52)	Examine perceptions of GD among people with a family history of HD	Qualitative survey analysis: in open-ended questions, participants requested to describe GD situations Narrative findings Content analysis	Stigma and GD seen as related concepts. GD: adverse treatment based on family history or genotype, without symptoms (Rothstein & Anderlik, 2001). Stigma: co-occurrence of components including labeling, stereotyping, separation, and loss of status (Link & Phelan, 2001). A key aspect is perception of an attribute of the stigmatized person conveying a devalued social identity (Crocker et al., 1998)	Core theme of Information Control developed, with themes of What They Encountered, What They Felt, What Others Did, What They Did Themselves, and What Happened Obstacles hinder access to goods and services usually available to healthy people, for example, organizational policies precluding insurance; job losses; unsupportive colleagues; poor treatment socially with friends and family posttest Emotional responses disclosed: overwhelmed by stigma and discrimination; sense of futility and injustice

Abbreviations: GD, genetic discrimination; HD, Huntington's disease; PHAROS, Prospective Huntington At-Risk Observational Study.

TABLE 4 Summary of quantitative study characteristics and key findings.

Author (year)	Country of origin	Sample (participants meeting criteria for review)	Aims	Methodology	Type(s) of stigma explored, including definition (if given)	Findings related to stigma
Boileau et al. (2020)	USA	479 people with a positive test and/or diagnosis of HD 192 premanifest 191 early stage 96 late stage 58.3% female 96% White, 1.9% African American, 2.9% other Age in years at baseline M (SD): 49.1 (13.1)	Explore which domains of health-related quality of life have greatest impact on perceived stigma in individuals with HD over time	Multilevel model used to explore effects of cognitive, emotional, physical, and social health, controlling for demographic factors, upon perceived stigma, assessed using Neuro-QoL Stigma at baseline, 12-month and 24-month follow-ups	Goffman's (1963) definition: stigma is an "attribute that is deeply discrediting" (p. 3), resulting in nonacceptance in society. Link and Phelan (2001): people perceived as different with perceived undesirable characteristics are placed in an out-group, resulting in a loss of social, economic or political power	Between-subject demographic variables (sex, race and ethnicity) not significantly associated with stigma Time, stage of HD and physical, emotional, social and cognitive health-related quality of life associated with perceived stigma, explaining 20.2% of variance
Bombard et al. (2009)	Canada	233 asymptomatic people: 83 tested positive for the HD gene expansion 84 tested negative 66 chose not to be tested 65.7% female Age in years M (SD): 45.5 (11.7)	Assess the nature and prevalence of GD within people at-risk of HD	Self-administered survey Cross-sectional	GD defined as differential treatment of asymptomatic people according to genetic differences rather than physical features (Billings et al., 1992)	GD reported by 39.9%, most frequently in insurance (29.2%), family (15.5%) and social (12.4%) settings, with fewer experiences within healthcare (8.6%), employment (6.9%) and public sector (3.9%) settings Family history of HD was the main cause of experiences of GD (rather than test outcomes) Experiencing GD associated with psychological distress ($p < 0.001$)
Bombard et al. (2011)	Canada	233 asymptomatic people: 83 tested positive for the HD gene expansion 84 tested negative 66 chose not to be tested 65.7% female Age in years M (SD): 45.5 (11.7)	Describe sociodemographic characteristics and factors associated with the experience of GD in people at-risk of HD	Self-administered survey Cross-sectional	GD defined as differential treatment of asymptomatic people according to genetic differences rather than physical features (Billings et al., 1992)	Greater likelihood of GD experiences across social, insurance and family contexts if: learned of being at-risk at a younger age (OR = 2.35; 95% CI: 1.13–4.86; $p = 0.022$); younger current age (OR = 0.97; 95% CI: 0.94–1.00; $p = 0.038$); positive genetic status (OR = 2.26; 95% CI:1.06–4.82; $p = 0.036$)

(Continues)

TABLE 4 (Continued)

Author (year)	Country of origin	Sample (participants meeting criteria for review)	Aims	Methodology	Type(s) of stigma explored, including definition (if given)	Findings related to stigma
Bombard et al. (2012)	Canada	233 asymptomatic people: 83 tested positive for the HD gene expansion 84 tested negative 66 chose not to be tested 65.7% female Age in years M (SD): 45.5 (11.7)	Explore nature and prevalence of concern for discrimination against oneself and family members; describe mitigating strategies; identify factors associated with experiences and concerns of GD	Self-administered survey Cross-sectional	GD defined as being treated unfairly or unfairly prevented from doing something (Bombard et al., 2008), and as differential treatment of asymptomatic people according to genetic differences rather than physical features (Billings et al., 1992)	86% of participants feared discrimination, particularly in insurance, family, and social contexts More concern about GD experiences among those with higher education, who learned their at-risk status when younger, and who were gene-positive Prevalence of overall GD concern for self almost 2× higher than prevalence of reported experiences of GD (i.e., concerns re. GD exceeded experiences)
Carlozzi et al. (2020)	USA	362 individuals: 152 premanifest 210 manifest HD (early HD $n=151$; late-HD $n=59$) Combined sample age in years at baseline M (SD): 49.1 (13.0) 58.6% female 96.1% White, 1.7% African American, 1.9% other, 0.3% not stated Data from HDQLIFE cohort	Determine reliability and responsiveness of Neuro-QoL and Patient-Reported Outcomes Measurement Information System (PROMIS) mental health patient-reported outcomes in relation to HD progression over 24 months	Longitudinal analysis of Neuro-QoL data at baseline, 12-month, 24-month follow-up One-way ANOVA comparing mean changes	No stated definition or concept of stigma; however, Neuro-QoL Stigma outcome measure described as assessing perceived self-discrimination	Differences between premanifest and both early HD and late HD groups for Neuro-QoL Stigma at baseline. Significant differences between all groups at 12- and 24-month follow-up Neuro-QoL and PROMIS mental health measures had strong psychometric reliability and responsiveness to self-reported and clinician-rated change over time
Carlozzi et al. (2019)	USA	294 individuals: 192 remanifest 192 manifest HD (early HD $n=131$; late-HD $n=61$) Combined sample age in years at baseline M(SD): 49.61 (12.84) 57.8% female 97.3% Caucasian, 2.4% other, 0.3% unknown	To establish psychometric properties of Neuro-QoL and PROMIS measures of psychological functioning	Cross-sectional analyses of reliability, floor/ceiling effects, timing data, convergent and discriminant validity, known-groups validity, effect sizes	No specific definition or concept of stigma described; however, Neuro-QoL Stigma outcome measure described as assessing perceived self-discrimination	Significant group differences between premanifest and early HD; premanifest and late-HD; early HD and late-HD – reported stigma increased as HD stage progressed Neuro-QoL Stigma Computer Adaptive Test M (SD): premanifest 45.19 (6.87); early HD 49.88 (8.46); late-HD 53.63 (8.54); Total sample 48.9 (8.5)

TABLE 4 (Continued)

Author (year)	Country of origin	Sample (participants meeting criteria for review)	Aims	Methodology	Type(s) of stigma explored, including definition (if given)	Findings related to stigma
Sokol et al. (2023)	USA	322 participants with HD gene expansion: 50 prodromal 101 early HD 101 late-HD Age in years M (SD): 51.6 (12.72) 46% female 95% Caucasian, 3% African American, 2% Other	Evaluate whether suicidal thoughts and behavior and HRQOL patient-reported outcomes contribute to advanced care planning and whether advanced care planning relates to changes in suicidal thoughts and behavior and HRQOL at 24 months	Longitudinal analysis of patient-reported outcomes related to HRQOL obtained at baseline, 12-month, 24-month follow-up Linear mixed-effects modeling of relationships between suicidal thoughts and behavior and HRQOL baseline and follow-ups	Not defined or described; Neuro-QoL Stigma used as patient-reported outcome	Scores for Neuro-QoL Stigma measure at baseline M (SD): 51.3 (8.78) Worsening of scores noted at follow-ups nonsignificant after correcting for multiple comparisons. Authors suggest people with HD who engage in advanced care planning may simultaneously engage in "grief work" (p. 84) while perceiving a loss of respect and independence, hence the trend toward increasing sense of stigma

Abbreviations: ANOVA, analysis of variance; GD, genetic discrimination; HD, Huntington's disease; HRQOL, health-related quality of life.

definitions were used more prominently, notably Goffman's work on stigma, with frequent references to people with HD or the gene expansion trying to manage "tainted" features of their identity and pass as "untainted." Participants referenced strategies of concealment across work, social, romantic, and organizational contexts, as well as self-isolation, appearing to bear out the relevance of Goffman's conceptualization of stigma in populations affected by HD. Of course, the model applied also potentially influences interpretation of data top-down (i.e., the preexisting model shaping the findings; for more on this, see the Discussion).

In terms of genetic discrimination, Billings et al.' (1992) conceptualization appeared to predominate, with multiple papers describing the differential treatment of people both at risk of HD and gene-positive but presymptomatic. Participants described both actual experiences of genetic discrimination and fear of those that might be encountered in future. Genetic discrimination was perceived as an actual or experienced risk across familial, insurance, employment, social, and healthcare contexts, with a particularly notable impact of experiences of other family members further along in the HD trajectory. This appeared to provide support for the relevance of Billings et al.'s conceptualization of genetic discrimination in HD contexts.

Crucially, half of the qualitative papers did not reference a theoretical definition of stigma either adequately or at all; nor did the vast majority of the quantitative studies or two-thirds of the mixed-methods papers. For genetic discrimination, no theoretical definition was provided for two-thirds of qualitative papers, over half of quantitative papers, and the vast majority of mixed-methods papers. Some papers did not focus with an adequate degree of specificity on genetic discrimination, with a broader focus which included visible symptoms and signs of HD progression or just exploring wider discrimination in general. Again, we will return to these limitations in the Discussion due to their significant implications, but the generally atheoretical position of a surprisingly large proportion of the literature provides important context for the remaining results and is therefore noted here for context.

3.2 | Methodological approaches utilized to explore stigma

3.2.1 | Designs

The 32 identified papers comprised 19 qualitative, seven quantitative, and six mixed-methods designs.

Qualitative designs included 14 individual interview papers, three using focus groups, one applying a qualitative analysis to narrative responses from open-ended questionnaire items, and one reporting brief case studies (see Table 3 for details). Seven of these specifically aimed to explore experiences related to genetic discrimination, and one aimed to explore perceptions of stigma. Of the former, five analyzed individual interview data via grounded theory, which seeks to develop theory derived from data (Charmaz, 2006), and two applied content analysis to analyze either interview data or narrative

TABLE 5 Summary of mixed-methods study characteristics and key findings.

Author (year)	Country of origin	Sample (participants meeting criteria for review)	Aims	Methodology	Type(s) of stigma explored, including definition (if given)	Findings related to stigma
Erwin et al. (2010)	USA, Canada, Australia	433 asymptomatic people at-risk for HD 71.8% female 67.8% from USA, 21.5% from Australia, 10.7% from Canada Age in years M (SD): 45.54 (10.65)	Explore GD prevalence; explore instances of adverse treatment and where knowing one's genetic risk status was beneficial in contexts of employment or insurance	Cross-sectional study: survey with open and closed questions about feelings, experiences or actions relevant to knowing family history of HD and own genetic test result	Defined GD as denial of rights, opportunities, privileges or other adverse treatment based solely on genetic information, including family history or genetic test results (Gostin, 1991) Stigma defined as co-occurrence of components including labeling, stereotyping, separation, and loss of status (Link & Phelan, 2001); primary feature is perception of an attribute conveying a devalued social identity within a particular social context (Crocker et al., 1998) Stigma and GD both described as the outwardly demonstrated aspects of a socially devalued identity existing beyond the ability of the person to determine	46.2% participants had experienced some form of unfair treatment; 56.5% reported >1 instance USA participants reported less GD and stigma (41.3%) than Canada (50%) and Australia (60.2%) GD instances categorized in four domains: (1) relationships (32.9% reported ≥1 instance), (2) insurance (25.9%), (3) employment (6.5%), and (4) GD or stigma in daily transactions (4.6%) Participants reported worry about possible GD at greater rates than experienced incidents; for example, 51.2% worry about GD or stigma in relationships, versus 32.9% reporting experience of GD in this context
Goh et al. (2013)	Australia	60 participants from one PREDICT-HD site: 36 gene-positive 24 gene-negative 65% female Age in years M (SD): 44.6 (12.4) These findings also comprise the Australian results of Erwin et al. (2010)	Examine GD among those clinically undiagnosed with HD	Cross-sectional survey; open and closed response formats exploring perceptions of GD	GD defined as differential or other adverse treatment, or denial of rights, privileges or opportunities, based solely on genetic information, for example, family history (Gostin, 1991) including if asymptomatic (Billings et al., 1992). Survey used terms "differential treatment," "treated unfairly," and "discrimination" in attempt to minimize response bias (Treloar et al., 2004)	27% expressed concerns about being treated differently following genetic testing; 17% strongly concerned about privacy of results 33% perceived instances of GD, which happened repeatedly causing great distress More gene-positive participants ($n=17$) reported experiencing GD, compared with three gene-negative participants ($\chi^2(3)=8.78$, $p=0.032$) GD occurred in the context of insurance (58%), then employment (21%), daily transactions (16%), and relationships (5%)

TABLE 5 (Continued)

Author (year)	Country of origin	Sample (participants meeting criteria for review)	Aims	Methodology	Type(s) of stigma explored, including definition (if given)	Findings related to stigma
Lemke (2009)	Germany, Austria, USA	48 people recruited via Deutsche Huntington-Hilfe (German HD self-help organization) quarterly publication Sample and eligibility not described; greater proportion of symptomatic people than people living at risk	Explore experiences of GD for people affected by HD	Questionnaires with both closed and open questions, followed by telephone interviews for respondents who indicated experiences of GD to gain further information. Nine interviews were conducted lasting between 12 min to over an hour	GD defined as a negative differential treatment on the basis of known or assumed attributes of a person's genetic makeup Used Goffman's (1963) theory of stigma to analyze results	60.4% reported experiences with GD; 18.8% reported fear of future GD; 20.8% reported neither Most who declared GD reported instances of stigmatization, disrespect and exclusion based on existing symptoms Four persons at-risk reported institutional GD, for example, instant rejection from health insurance companies due to family history of HD Participants concealed their genetic status or gave partial information (e.g., in job applications) to avoid GD: "precautionary secrecy" (p. 27) Discriminatory treatment from medical professionals (e.g., physician suggesting sterilization)
Taylor (2005)	Australia	57 participants: 45 at 50% risk 5 at 25% risk 7 had undertaken the predictive test 66.7% female. Age in years M (SD): 38.8 (12.9)	Explore gender differences in attitudes of people at-risk for HD regarding predictive testing and genetic risk	Cross-sectional postal survey exploring genetic status, beliefs, and attitudes around at-risk status and predictive testing Thematic analysis of open-ended question responses Chi-square significance testing and logistic regression of predictors of testing intentions	Stigma not described or defined; "discrimination" discussed in results and discussion	Majority had not disclosed to employers 89% reported their family history or genetic status had not resulted in discriminatory treatment; 54% felt vulnerable and feared discrimination Males 3× more likely to fear disadvantage than women (Fisher's exact test, $p=0.03$). Authors suggest this may influence the larger negative inclination of men toward testing and could impact participation in testing and research

(Continues)

TABLE 5 (Continued)

Author (year)	Country of origin	Sample (participants meeting criteria for review)	Aims	Methodology	Type(s) of stigma explored, including definition (if given)	Findings related to stigma
Thorley et al. (2018)	USA	115 participants: 80 with HD 35 caregivers supporting a person with HD 75% female Mean age 45 years 99% white; 1% mixed race	Evaluate how chorea impacts HRQOL and overall functioning among people with HD	Cross-sectional survey comprising open and multiple-choice questions Patient-reported Neuro-QoL Stigma scores compared between those experiencing high versus low chorea using multivariable linear regression model	Not explicitly stated; Neuro-QoL Stigma measure described as assessing patient perceptions of stigma	Significantly higher Neuro-QoL Stigma scores among those experiencing higher chorea than lower chorea [mean (SE): 60.9 (1.0) versus 50.5 (1.3); $p \leq 0.0001$] Positive linear association between stigma and chorea ($r = 0.66$, $p < 0.0001$) suggesting perception of greater social stigma was associated with a higher impact of chorea on HRQOL In regression model, as Neuro-QoL Stigma scores increased (indicating higher perceived stigma) chorea scores also increased ($p = 0.0031$)
Velissaris et al. (2023)	Australia	7 people presymptomatic for HD 71.43% women Mean age in years: 49, ranging from 36 to 60	Pilot study evaluating whether an 8-week mindfulness-based stress reduction program for people presymptomatic for HD was feasible and acceptable, embedded an understanding of mindfulness, and had potential to reduce distress and enhance psychological adjustment	Quantitative repeated measures pre- and post-intervention Qualitative data collected via semi-structured interviews post-program	Stigma/social stigma, not defined; acknowledgment that people with presymptomatic HD will usually have experienced social stigma	Qualitative feedback suggested program reduced sense of isolation and stigma, and allowed positive connection related to HD: "Meeting other people with HD is a huge thing…they know exactly how you're feeling" (p. 6); "[The course] de-isolated us" (p. 6); "In this group, HD is not my dirty little secret" (p. 6); "Would have been great to have the opportunity to talk more because in my family, it was not talked about …it is nothing to be ashamed of" (p. 6)

Abbreviations: GD, genetic discrimination; HD, Huntington's disease; HRQOL, health-related quality of life; PREDICT-HD, the Neurobiological Predictors of Huntington's Disease study.

survey responses to reveal patterns of themes and the magnitude of concerns reported. Etchegary (2007) used interpretative phenomenological analysis to explore how participants made sense of stigma in the context of their lives. The remaining six qualitative papers did not explicitly aim to explore stigma or associated constructs, but aspects relevant to stigma were reported (hence their inclusion in this review).

Seven papers reported quantitative methods (see Table 4), comprising three longitudinal and four cross-sectional studies. Three papers from Bombard et al. (2009, 2011, 2012) specifically aimed to explore genetic discrimination via cross-sectional surveys developed from qualitative work, questions adapted from validated instruments, and input from an expert reference group. Only one study (Boileau et al., 2020) explicitly aimed to explore multiple domains influencing perceived stigma, using a self-reported, previously validated measure of perceived stigma (Neuro-QoL Stigma; Cella et al., 2012) as an outcome variable. Another study used Neuro-QoL Stigma as both predictor and outcome (Sokol et al., 2023) to evaluate whether health-related quality of life variables were associated with future advance care planning over 24 months. Two articles concerned psychometric properties of patient-reported outcome measures, via cross-sectional and longitudinal designs (Carlozzi et al., 2019, 2020). Three papers (based on two samples) demonstrated significant group differences across stages of HD, suggesting perceptions of stigma increased with HD progression over 2 years (Boileau et al., 2020; Carlozzi et al., 2019, 2020).

Finally, six papers described a mixed-methods approach (see Table 5); five reported cross-sectional surveys that gathered data via open and closed questions with a descriptive qualitative design (one study followed the questionnaire with additional individual interviews; Lemke, 2009). The remaining paper used a repeated measures experimental design (Velissaris et al., 2023). Two papers did not explore stigma via both methods; one reported a sense of reduced isolation and stigma which only emerged within post-intervention feedback interviews (Velissaris et al., 2023), while another study only examined perceived stigma using Neuro-QoL Stigma (Thorley et al., 2018). Three articles (Erwin et al., 2010; Goh et al., 2013; Lemke, 2009) aimed to explore experiences of genetic discrimination across open and closed questions; Taylor (2005) also employed a similar survey which produced results relevant to discriminatory treatment. Three studies did not present a qualitative analysis of responses to open-ended questions (Erwin et al., 2010; Goh et al., 2013; Taylor, 2005); however, the former two stated that this is reported in Williams et al. (2010), also included in this review.

In sum, there was a broad disparity in approaches to exploring stigma and genetic discrimination among the identified papers. Just over half of the qualitative papers intentionally explored experiences related to genetic discrimination or perceptions of stigma, meaning that for the remainder, findings relevant to these constructs appear to have been obtained in a relatively unintentional or unstructured way, and therefore cannot be assumed to have explored the subjects in full (although their content is nonetheless highly relevant to this review). For the quantitative methods papers, the disparity of underpinning definitions appears to feed through to a range of approaches to measurement and analysis, with varied tools and analytic methods used to approach understanding these constructs through a diversity of methodological approaches. This is also the case for the mixed-methods papers, where the predominant approach was cross-sectional surveys with a range of open and closed questions. Again, there is a paucity of theoretically driven coherence across the field at present, and we will return to the implications of this in the Discussion.

3.3 | Gaps and areas of underrepresentation

In this section, we will note gaps or issues in the set of identified papers, but will defer analysis and interpretation of the impact of these issues until the Discussion.

Most studies recruited via specialist clinics for HD and/or neurological disorders, thus accessing participants in receipt of medical care related to HD, generally using convenience sampling. Only one study (Boileau et al., 2020) investigated the impacts of other characteristics (beyond HD status) for which people may already experience stigma within society, such as gender, age, sexuality, and ethnicity. While Boileau and colleagues' findings suggested that age, race, and ethnicity were not significantly associated with stigma, they acknowledged that their sample was primarily white and highly educated, which limited their ability to effectively investigate these questions among their sample. Most papers suffered from similar sampling bias around lack of diversity, with samples being predominantly white, educated women.

Some papers commented on the cultural context in which people experienced stigma; however, many did not, despite this being essential information to consider given the contextual experience of stigma (Crocker et al., 1998). Furthermore, a significant global north bias was evident across the included studies. There were no included studies from African countries, and only one study each from Asia and South America. The dearth of articles from the global south may be partially explained by inclusion being limited to articles in English due to the lack of resources for translation. A further under-recognized problem is the systematic disparity of representation of research from the global south more widely, with such articles being less likely to be indexed, often published in lower prestige journals, and overall less likely to be identified in context of reviews (Bol et al., 2023).

Considering eligibility criteria among selected papers, 15 papers sampled only at-risk, asymptomatic participants, including family members not at-risk and/or those receiving negative test results. Additionally, participants below 18 years old were excluded in most studies bar Kjoelaas et al. (2022), indicating an underrepresentation of views from younger people. Many papers did not acknowledge the potential for differences across experiences of stigma depending on the specific HD population, with participants with manifest HD compared with presymptomatic and at-risk individuals. All three populations are likely to experience stigma and genetic

discrimination in different but overlapping ways, ranging from reactions to visible signs of HD (for manifest groups, though arguably this could meaningfully be broken down even further by HD stage/severity of progression), confirmed future development of HD (gene-positive presymptomatic groups), or those at risk of possibly developing HD in future.

Finally, 16 papers did not offer clear recommendations for how to address stigma in their discussion or conclusions. Five suggested individual interventions, such as counseling, to reduce associated distress. Bombard et al. (2009) and Boileau et al. (2020) specifically referred to targeting "mental health," potentially suggesting the positioning of stigma as more a subjective, individual variable as opposed to a societal problem. Four papers suggested legal avenues including policy changes, advocacy, and advice (Bombard et al., 2007; Penziner et al., 2008; Taylor, 2005; Wauters & Van Hoyweghen, 2018). Six advised healthcare professionals to provide information about stigma and genetic discrimination with the aim of mitigating its impact, and to provide signposting for additional psychosocial support (e.g., Etchegary, 2007; Wauters & Van Hoyweghen, 2021). The most prominent recommendation, suggested in 10 papers, was for societal interventions to raise public awareness of HD, with the aim of facilitating greater understanding and compassion, and reducing stigma and genetic discrimination (e.g., Boileau et al., 2020; Bombard et al., 2011; Leung & Leung, 2002). These recommendations provide potential routes to clinical and social impact around these difficulties faced by people affected by HD, although the significant disparities and limitations of the existing evidence base must be taken into account when considering such steps.

4 | DISCUSSION

4.1 | Summary

This scoping review aimed to explore key concepts, definitions, methodological approaches, and gaps and areas of underrepresentation across studies describing experiences of stigma among people affected by HD. Thirty-two papers were included, although several used the same samples or overlapped participants between included papers.

While the search specified papers published between 1993 and 2023, those included were published between 2002 and 2023, indicating rising interest in investigating experiences of stigma in more recent years. For instance, within the 30-year inclusion period, five articles were published in the first 15 years while 27 papers meeting eligibility criteria were published from 2008 onwards. This rising recent interest likely relates to increasing attention to HD mental well-being and growing acceptance of more systemic understandings of psychological difficulties among HD families (e.g., Lindo et al., 2026), following a long-term predominance of clinical and academic focus on physical and motor difficulties associated with HD. However, the majority of papers did not report specific aims addressing stigma or associated constructs, suggesting that despite the trend in publications relevant to stigma over time, experiences of stigma were identified incidentally or, at best, were complementary to the papers' research questions.

4.1.1 | Key concepts and definitions of stigma in HD research

Almost half of papers did not provide a conceptualization or established theoretical understanding of stigma. Underpinning constructs such as felt/enacted stigma (Scambler, 1998) were rarely discussed in HD literature, suggesting a theoretical gap in which some papers appeared to report experiences of felt/enacted stigma without identifying it as such. This omission could suggest a lack of theoretical grounding, whereby researchers circle around these key constructs while conducting somewhat atheoretical research in terms of conceptualizing stigma. This may signal a problematic lack of conceptual underpinnings in the research, given that theoretical frameworks distinguishing constructs related to stigma were not offered. In other cases, theoretical frameworks and definitions were referenced as part of establishing context in paper introductions but then only re-referenced in the discussion sections or not at all, again suggesting a lack of robust theoretical integration driving empirical work. Unfortunately, this lack of definitional clarity is not confined to HD research. While problematic in its own right, this also presents a barrier to coherent and actionable research (see Thomas, 2024, for a wider discussion on the unclarities and assumptions regarding the role of stigma in genetics in general).

This absence of clear theoretical grounding also presents difficulties in terms of understanding the position of the authors and its influences on analysis, particularly in the qualitative papers. There was a degree of consistency around definitions of genetic discrimination (Billings et al., 1992) and, to a lesser extent, stigma (Goffman, 1963). A surprisingly large number of papers either reported definitions not focused on the relevant constructs to an adequate degree, or reported no definition at all. It is difficult to see how a robust and theoretically motivated understanding of stigma or genetic discrimination can meaningfully be developed with this crucial information missing. It must, however, also be considered that any adopted conceptualization will inevitably exert top-down influence on study findings, shaping the data collected and its interpretation. Accordingly, it is essential that research into stigma and genetic discrimination in HD adopts a clear framework with a robust rationale for the choice, and direct communication of authors' positioning.

This is particularly important given recognized limitations, even where a relevant conceptualization was applied. For example, Goffman's (1963) discussion of stigma has been criticized for failing to acknowledge that "axes of risks for stigma" (e.g., characteristics around which people may already experience stigma, such as ethnicity, gender, and/or age) rarely present in isolation (Brewis & Wutich, 2019, p. 185); this is often referred to as the "intersectionality" of stigma and will be discussed further in context of gaps and underrepresentation below.

4.1.2 | Methodological approaches utilized to explore stigma

The identified papers used a range of qualitative, quantitative, and mixed-methods approaches. Longitudinal studies were rare. However, the recruitment strategies used (i.e., recruitment from hospitals and tertiary centers) could have omitted people affected by HD who were not in contact with medical support, such as those who choose not to be tested or who avoid engaging with services (e.g., for reasons relating to anticipated stigma, or practical problems impeding access). Many papers reported using convenience sampling, which carries many potential biases (although there are challenges associated with adopting probability sampling methods in populations with rare conditions). Self-selected samples might limit generalizability to wider populations affected by HD, as individuals who might be more sensitive to stigma, or those who engage less (or not at all) with the HD community, may be less well represented in the findings, especially where studies recruited via a single source or site. Further issues relating to sampling bias will be discussed in the subsequent section.

Methodologically, it is crucial to consider the choice of population. Some identified studies did not adequately distinguish between people who are at-risk, presymptomatic or manifest, which incurs potential limitations. A person with HD who has visible chorea, problems with mobility, involuntary facial movements and speech difficulties is likely to encounter stigma in relation to their visible symptoms, as borne out in our review findings (a common example from clinical experience is a person being considered drunk due to unsteady walking). A person who is gene-positive for HD and presymptomatic may, based on our findings, encounter genetic discrimination (e.g., barriers to employment/promotion), but does not possess these sorts of visible differences and therefore others' responses may differ on that basis. A person who is at-risk may encounter different forms again (e.g., judgment around choices to have children or not). Clinical and lived experience tells us that these are likely to be experienced as very different situations, but the research base presently does not provide evidence to confirm or deny, and this accordingly should also be considered in any related research going forward.

4.1.3 | Gaps and areas of underrepresentation

Several important gaps and areas of underrepresentation in the literature around HD stigma are apparent, in particular the lack of studies with young people and from countries not part of the global north. In terms of the variables or experiences explored, the relative lack of intersectionality was noted. "Intersectional" experiences of stigma may accumulate across axes of identities, differentially impacting people of varied oppressed identities (including that of living with HD) and potentially increasing vulnerability to labeled differences associated with negative stereotypes, further separating them from others without such identities (Link & Phelan, 2001; Tyler, 2020). Thus, it is imperative to deliberately seek experiences from those often marginalized, and include these perspectives so that the findings are applicable to typically excluded and underrepresented groups.

It is particularly important to consider cultural differences in stigma around physical health difficulties and genetic conditions, an area also underrepresented in the wider rare disease literature (Baynam et al., 2024). With samples in the reviewed papers predominantly comprising white, educated women, and the generally sparse literature on stigma and HD, there is presently very little understanding of how stigma and genetic discrimination may impact on members of minoritized groups in these contexts. Most of the papers reviewed here only reported gender and age, and there was extremely limited discussion as to how different forms of stigma can cause cumulative harm and reinforce social and health inequalities (Hatzenbuehler et al., 2013), linking back to concerns regarding how well these issues are even represented in the current widely used models of stigma. For example, Goffman (1963) does not explicitly address environmental and structural forces shaping disability and did not recognize how personal interactions are influenced by social, economic, and political power (Gleeson, 1999), contrary to the later-developed social model of disability (Oliver, 1996). This presents a significant omission when attempting to understand stigma as an inherently interpersonal and social phenomenon.

This not only represents a theoretical oversight, but neglects the already under-acknowledged needs of minoritized and vulnerable groups. Tyler (2020) considered stigma as a form of power and violence, condemning passive psychological conceptualizations focusing on individual experiences without exploring its structural power to cause social and political injustice. Therefore, researchers must resist framing stigma as an issue within the individual, or interpreting a pronounced sense of discrimination as a faulty cognition indicative of disordered thinking requiring "treatment" (Simpson & Thomas, 2015). Researchers should instead consider stigmatization as an ongoing process and tool of oppression examined in interactional (micro) and structural (macro) approaches (Thomas, 2021) in the pursuit of social justice. This argument did not feature within selected papers, although has been suggested in relation to stigma around Parkinson's disease (Simpson et al., 2013), and we would argue for it as an essential perspective in understanding and reducing stigma going forward.

4.2 | Strengths and limitations

The present scoping review was strengthened by broad inclusion criteria to capture a wider range of papers including people affected by HD. Consequently, this review includes a wide scope of how stigma has been researched and conceptualized, offering insight into areas of underrepresentation. A significant limitation of this review is that both the review authors and the majority of the included studies were situated in the global north, reflecting

a dominance of northern epistemologies that may marginalize perspectives, priorities, and knowledge systems from the global south, thereby limiting the inclusivity and global relevance of the findings.

Papers published before 1993 were excluded, yet untested participants at-risk of HD were still included. Acknowledging this limitation of a broad and inclusive scoping approach, the rationale was to focus on papers produced following the introduction of accessible genetic testing which has been associated with experiences related to stigma (e.g., Bombard et al., 2012). Finally, experiences from those without a genetic history of HD were excluded (e.g., spouses/caregivers). While this enabled a focus on experiences of people with the gene expansion for HD, research has demonstrated that caregivers were more likely to report experiences of enacted stigma (Etchegary, 2007), which could indicate a potential avenue for future reviews.

4.3 | Directions for future research

It is clear that future research would benefit from incorporating a more explicit and in-depth conceptualization of stigma. Of the various frameworks included to date, Scambler's (1998, 2004) work is particularly relevant to people with HD given its focus on the different forms of felt and enacted stigma. While Goffman was influential in the development of Scambler's approach, and continues to be cited in terms of definitions and the concept of visible and invisible identities, Scambler's work captures both the *social reality of discrimination* and the *internal, anticipatory, and emotional dimensions* of stigma—all of which appear central, albeit at different points, to the HD experience. This approach can also be linked with other theoretical frameworks, for example, Link and Phelan (2001) in relation to structure, or Earnshaw and Chaudoir (2009) regarding the mechanisms of stigma.

Most of the identified papers presented cross-sectional research, precluding exploration of how perceptions of stigma may change over time such as in relation to HD progression. Therefore, longitudinal experimental designs recruiting from diverse populations affected by HD may improve validity, reliability and hence the utility of findings in relation to drawing meaningful conclusions about associations between other variables (e.g., societal attitudes, genetic status, mobility, and other visible differences). Future research (both qualitative and quantitative) would also benefit from analyses investigating and comparing different groups affected by HD, clearly delineating those at-risk, presymptomatic and manifest, to explore whether experiences of stigma differ between groups.

Moreover, only five papers used a validated quantitative measure of stigma, while others (e.g., Bombard et al., 2009, 2012) used a nonvalidated survey. Use of a range of nonvalidated measures may impede the effectiveness of exploring stigma experienced by people with HD; future research could seek consensus regarding how best to evaluate perceived stigma, such as surveying clinical professionals and experts-by-experience as panelists to develop appropriate criteria via a Delphi study (Hardy et al., 2004; Nasa et al., 2021). Such assessments could subsequently be used within routine appointments at HD clinics, for instance, psychosocial assessment of functioning and participation within healthcare to facilitate access to social support if indicated. In other related conditions, such as Parkinson's, bespoke tools have been created which are theoretically informed (e.g., Stopic et al., 2024).

Finally, exploring stigma at the societal level could be a valuable future direction, and qualitative research methods may make a particularly important contribution in this area. For example, discourse analysis of stigmatizing messages about health and genetic disease in a culture's media may highlight normative scripts othering people affected by genetic disease, such as portraying assumptions of a life of tragedy and hardship (e.g., Thomas, 2021). Such discourses might increase fears of judgment, and the stigma associated with HD, such as assumptions about disability or the burden of caregiving, might discourage individuals from seeking genetic counseling and predictive testing.

4.4 | Clinical implications

For genetic testing professionals, while a comprehensive process is often in place, *explicit discussion of the different types of stigma* may not always be systematically standardized (Baynam et al., 2024). Given the nature of HD, it would make sense for genetic counseling protocols explicitly to include the different forms of stigma, for example, anticipated stigma (including fear of discrimination), enacted stigma possibilities (regarding insurance, employment, family reactions) and internalized stigma (how individuals might feel about themselves and their identity). Moreover, this discussion should be revisited as clearly different scenarios and challenges may manifest at different points throughout the genetic testing process (and beyond). Genetic counselors can also refer to support organizations which can play a role in modifying stigma in people with neurological conditions (e.g., Elafros et al., 2013).

A clear omission in the current evidence base is any intervention paper seeking to reduce stigma (or its effects) for people affected by HD (Zarotti et al., 2020). Despite its harmful effects being outlined repeatedly (e.g., Lancet Editorial, 2010), limited suggestions have been made as to how to address stigma and enact change. While public information campaigns (e.g., Lancet Editorial, 2010) have their place, multilevel actions plans are needed (e.g., Rao et al., 2019). Such levels can be intrapersonal/individual (aimed at addressing "burden" narratives); interpersonal and community-focused (aimed at schools/ workplaces); organizational and institutional (aimed at healthcare professionals/social care employees); and structural/ society level (aimed at legislative, wider policy change). Moreover, careful development—involving individuals and families affected by HD—and evaluation is needed to avoid unintended consequences, such as reinforcing stereotypes (Thomas, 2024). While admittedly challenging and hard to implement, only when such comprehensive

anti-stigma campaigns are in place can people affected by HD begin to see some relief from discrimination, and gain increased access to support and greater acceptance within society.

5 | CONCLUSION

In conclusion, this review has highlighted that, while research interest in the effects of stigma among people affected by HD has increased, the field is still in need of theoretical cohesion and development. The effects of stigma on the well-being of people affected by HD seem clear; routes to change that, however, remain less well defined and a strategic, theoretically informed multilevel approach needs to be developed and tested as a priority. However, we consider that only when the field is more theoretically developed can (1) specific HD-relevant tools be created to meaningfully measure stigma and (2) interventions be co-developed and tested to mitigate and reduce its effects.

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Tierney Tindall: Conceptualization; methodology; formal analysis; writing – original draft; writing – review and editing. **Jane Simpson:** Conceptualization; methodology; writing – review and editing; supervision. **Maria Dale:** Conceptualization; methodology; writing – review and editing; supervision. **Sarah Gunn:** Conceptualization; methodology; writing – review and editing; supervision.

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SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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