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Do Influential Articles on the Genetics of Autism Show Evidence of Engagement With the Autistic Community?

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ABSTRACT

Investigations into the etiology and genetic basis of autism continue to drive much autism research, yet reports are emerging of this research not aligning with priorities of autistic people. Engagement of autistic people in the research process is a key way to take their perspectives on board. We investigated whether influential genetic autism research shows evidence of engagement with the autistic community via indicators in published article texts. Through text mining of the abstracts of articles mentioning the words "autism" or "autistic," we found minimal prevalence of progressive terminology associated with autism. We also devised a novel rating system to assess three hallmarks of autistic community engagement: presence of non-stigmatizing language, referencing community priorities, and the use of participatory methods. We reviewed 149 articles within leading autism and genetic journals. Minimal evidence of engagement with the autistic community was found within all three hallmarks. Genetics researchers focused on autism should embrace opportunities to engage with the autistic community to bring their work into closer alignment with their priorities, yielding scientific and moral benefits.

1 | Introduction

1.1 | Autism and the Autistic Community

Autism is defined by lifelong distinctive social communication and interaction profiles, and the presence of restricted, repetitive, and inflexible behaviors (American Psychiatric Association 2013). Although officially listed as autism spectrum disorder (ASD) in the DSM-5, autism and autistic person (rather than person with autism) is the generally preferred word choice of the autistic community speaking English (Bottema-Beutel et al. 2021). Both environmental and genetic factors are believed to contribute to the occurrence of autism, yet a single cause or biological marker has not been uncovered, and hence diagnosis still relies on subjective assessments of behavior. There are some single-gene syndromes, such as fragile X syndrome or Angelman syndrome, which are strongly associated with autism; but even in these cases, behavioral testing must be used to confirm that autism diagnostic criteria are met. Additionally, although many of the genes implicated in these syndromes share molecular pathways, clinical presentation both between and within syndromes is highly heterogeneous (Parenti et al. 2020). Moreover, estimates of the overlap between syndrome and autism vary widely (e.g., Belmonte and Bourgeron 2006), and some have argued that there are important distinctions between the manifestation of autism in syndromic and non-syndromic cases (e.g., Abbeduto et al. 2014). The reliance on behavioral diagnostic assessment can pose a problem as the timing of an autism diagnosis plays a role in access to support systems, and people who have missed an early diagnosis are shown to have worse mental health and quality of life (Atherton et al. 2021)—though we also note reporting on risks as well as benefits of early diagnosis (Okoye et al. 2023).

Both the autistic community (comprised of autistic people themselves) and the broader autism community (further including their families, caretakers, and other stakeholders) advocate on behalf of autistic individuals and attempt to amplify autistic viewpoints. This includes facilitating direct advocacy by autistic people with a concurrent intellectual disability or language delay, and also advocating on their behalf from a shared autistic perspective and/or as a parent. There is a growing consensus that autism research should be influenced by the perspectives of the autistic and autism communities (Milton 2014; Fletcher-Watson et al. 2019). Patient and public involvement and engagement (PPIE) focuses on strengthening the relevance, quality, validity, and utility of research by emphasizing meaningful forms of power-sharing between researchers and the public (Russell et al. 2020). Informing the public about the processes or results of research is valuable, but retains all the power in the hands of the researchers: They decide what to share, with whom, and when, and the public can ask questions but with no guarantee of influence over how work is done in the future. Such power imbalances can be avoided with partnerships and delegated power, which provide a higher degree of citizen control (Arnstein 1969) allowing end-users (in this case, autistic people and their allies) to influence the work that is done. PPIE techniques and theory can be used to engage with autistic perspectives and include them in research, especially as autistic people want increased engagement from researchers (Pellicano et al. 2014b; Fletcher-Watson et al. 2019).

1.2 | Autism Genetic Literature

Autism genetic researchers are on a quest to find biological explanations for autism, and this research can potentially have a clinical impact. As the first twin studies that linked autism to hereditary factors (Folstein and Rutter 1977), estimates for autism heritability range from 40%–80% (Chaste and Leboyer 2022). It has been suggested that additive polygenic factors are responsible for the broader autism phenotype, which includes subtle differences in social communication, cognition, and executive functioning. This makes the autism genetic field highly desirable for researchers to make advancements in understanding the etiology of autism (Arnett et al. 2019). Studies have identified new loci that contribute to autism (Satterstrom et al. 2020), as well as elucidated genetic correlations between major depression and educational attainment with different clinical manifestations of autism (Grove et al. 2019).

Autistic people are frequently diagnosed with co-occurring conditions including epilepsy (Liu et al. 2022), congenital heart disease (Gu et al. 2023) and joint hypermobility (including Ehlers–Danlos syndrome; Baeza-Velasco et al. 2015). Understanding these associations at a genetic level may be beneficial to develop effective therapeutics. The impact genetic researchers strive for is to progress clinical care for autistic people by various efforts such as identifying genetic subtypes that would improve precision diagnostic and intervention methods (Arnett et al. 2019), translation into meaningful biomarkers that can aid in diagnosis as well as monitoring progress (Jeste et al. 2015) and to predict and improve outcomes (Carter and Scherer 2013). Genetic insights have the potential not only to impact scientific and clinical conceptualizations of autism and enhance understanding of the underlying neurobiology of autism but also to advance improvements to the health and quality of life of autistic individuals (Vorstman et al. 2017).

1.3 | Concerns Raised by the Autistic Community

Despite good intentions and potential for useful insights to be generated by genetic autism research, many members of the autism community have raised concerns regarding this research and its potential applications. There is widespread fear that advancements in understanding the genetics of autism could result in the development of prenatal testing and a pathway to termination, posing an existential threat to autistic people; this fear is not unfounded and already occurs in other neurodevelopmental conditions such as Down syndrome. Other concerns include worries about sharing and publishing data, and future misuse in a changing political landscape. Importantly, the choice by a relatively small number of autistic people to share their genetic data could have consequences for the entire population, so simple individual participant consent does not resolve the issues. These concerns are articulated in the substantial and well-organized negative responses from the autistic community in relation to one specific research announcement (Sanderson 2021; Natri 2021) but certainly apply to the field more generally (Natri et al. 2023).

This backdrop has generated a major gulf between autistic community leaders and genetics researchers. Disengagement from the autism community creates a significant problem, as failure to take action in response to community protest can contribute to their mistrust in researchers and an ineffective scientific process. As such, it is of paramount importance for autism genetics researchers to make strong connections with autistic people and their allies. In the context of a wider drive to include autistic perspectives in research generally, it is important to inquire whether this is occurring in autism genetic research, which also accounts for a large proportion of autism research funding and activity (Pellicano et al. 2013). If engagement is low, this opens up an obvious way to start to lessen the gap between community and researchers (noting of course that some people belong in both categories) by working with autistic people to do genetic research they find trustworthy and useful.

1.4 | Three Hallmarks of Engagement

A suitable first step toward building connections between genetics researchers and the autistic community is to examine the extent of community engagement to date. To do so, we identified three hallmarks of engagement that can be obtained from published literature: Use of non-stigmatizing language (Bottema-Beutel et al. 2021), research that explicitly mentions or cites community priorities (Pellicano et al. 2013, 2014b) and use of participatory methods (Nicolaidis et al. 2011; Fletcher-Watson et al. 2019).

Non-stigmatizing language can be defined as language that does not contribute to stigmatization and increased marginalization of autistic individuals (Bottema-Beutel et al. 2021). Identity-first language ("autistic person") is often considered

less stigmatizing than person-first formulations ("person with autism") (Gernsbacher 2017), but this is not consistent internationally (Buijsman et al. 2023; Keating et al. 2023). In addition, all attempts to capture a consistent preference between these two options show variability even amongst autistic people, and certainly when including the wider autism community (Kapp et al. 2013; Bury et al. 2023). However, there are forms of language which offer greater clarity. Examples of stigmatizing language include "burden," "suffering," "epidemic" and "cure," or discussions of the "risk" of autism, and these are common in research about autism (Gernsbacher et al. 2005; Kenny et al. 2016; Bottema-Beutel et al. 2021). Similarly, terms including neurodiverse or neurodiversity (referring to the natural diversity in human neurocognitive functioning) and neurodivergent (human neurocognitive functioning that diverges from dominant societal standards) (Walker 2014) can be used as indicators of positive engagement, because they represent consideration of autistic-led ways of theorizing about autism.

Despite the absence of universal and categorical delineations between stigmatizing versus progressive language, the choice of language is a useful marker for engagement, because it is highly visible in published work. Although stigmatizing language might be retained, for example, because of adherence to outdated scientific reporting norms, it is fair to assume that authors who make the choice to avoid stigmatizing language in their work will have engaged with autistic people. Language has the further advantage that it can be captured via automated text mining processes, permitting analysis of large numbers of publications at scale. Another hallmark that displays engagement with the autistic community is visible consideration of the community's priorities. There are a number of published accounts of autistic and autism community priorities for research (Cusack and Sterry 2016; Roche et al. 2021; Cage et al. 2024). Several studies have noted a difference in priorities between funded research and the autistic community (Nicolaidis et al. 2011), so engagement with the autistic community should lead to research that adopts autistic priorities (Pellicano et al. 2013). This should be visible in published work via explicit citation of community priority-setting reports and reference to community priority areas when describing the motivation and/or long-term implications of the study being reported.

The final hallmark of engagement is authors reporting the actual engagement itself, whether informal or formalized into participatory methods. Participatory methods and partnerships that foster inclusivity in research are considered best practices for conducting autism research (Nicolaidis et al. 2019). Participatory methods vary in the degree of shared power, and intend to "disrupt the power imbalance between the researcher and the participant" (Fletcher-Watson et al. 2019). Practices that fall under participatory methods include co-production, community-based participatory research (CBPR), consultation and citizen science (Fletcher-Watson et al. 2021).

1.5 | Aims and Hypothesis

The aim of this study was to discover whether influential autism and genetics papers show evidence of engagement with the autistic community. We ask, do papers on autism and genetics, published in leading autism and genetic journals, show evidence of:

- Using non-stigmatizing language?
- Considering autistic community priorities?
- Reporting informal or formal engagement directly with autistic people or their supporters?

We used two methods to address our question. First, we used automated textual analysis of a large corpus of autism research articles to explore the prevalence of non-stigmatizing language over time, and comparing across papers reporting in genetics journals versus others. We then used a constrained but systematic literature search, to identify influential autism genetics papers, and applied a novel rating scheme inspired by existing "risk of bias" evaluation tools (e.g., Sterne et al. 2016) to address all three questions.

According to previous literature, autistic people tend to want more research outside the genetic, biological, and cognitive domains (Pellicano et al. 2014b). But to our knowledge, there have been no attempts to quantify the extent to which research within these disciplinary categories (which includes genetic autism research) has engaged with the autistic community. Therefore, this project is necessarily exploratory, though we tentatively hypothesize, based on recent literature and discourse, that we will find little evidence of engagement as measured by our rating system.

2 | Methods

2.1 | Identifying Influential Journals

A list of the 10 most influential journals within autism and/or genetics was identified to achieve a manageable volume of articles for rating by hand (Table 1). "Influence" was operationalized with reference to various metrics including SCImago Journal Rank (SJR), impact factor (IF), h-index, and a total citation score above 1000 from the past 3 years. All the selected journals belonged to the 1st Journal Citation Reports (JCR) quartile, which is considered to encompass the highest quality scientific publications in the JCR evaluation system. The SJR accounts for the number of citations received by the journal, as well as the reputation of the source the citations come from. The IF is a ratio of the number of citations for a journal divided by the number of total citable items published by the journal over the past 2 years and is meant to reflect a rigorous review process. Despite IF being a dynamic metric that lacks clarity of what constitutes a good ranking, it is still deemed an appropriate indicator of a journal's reliability (Kaldas et al. 2020). H-index is a metric evaluating the cumulative impact of each individual author and the performance of publications, but it can be influenced by self-citation and biased toward more senior researchers. As such, the higher ranked journals in the SJR and IF metrics determined which journals were included in the list. Exceptions were highly ranked review journals, as our intention was to focus on the reporting of new data.

TABLE 1 | Selected journals for systematic literature search.

Journal title	SJR	IF	H-index	Total citation (2019–2022)
Nature Genetics	18.861	38.33	573	17,158
Genome Research	9.556	9.043	297	6386
Genome Biology	9.027	13.583	248	9017
Nucleic Acids Research	9.008	16.971	537	60,949
Cell Stem Cell	8.860	24.633	248	7913
Molecular Autism	2.638	7.509	56	3691
Journal of Autism and Developmental Disorders	1.374	4.291	175	12,223
Autism	1.899	5.689	96	4832
Autism Research	1.656	5.216	66	2480
Journal of Neurodevelopmental Disorders	1.431	4.025	45	1338

Note: List of selected journals with various metrics (values collected in April 2022). Data retrieved from (SJR: Scientific Journal Rankings, SJR 2022).
Abbreviations: H-index, Hirsch index; IF, impact factor; SJR, SCImago Journal Ranking.

2.2 | Text Analysis of Language Across Autism Publications

We conducted a broad search of the Web of Science (WoS) Core Collection database on January, Monday 15 2024 using the search term "TS=autism OR autistic OR ASD" to identify all indexed studies mentioning autism. We did not limit search results by article type or language; however, we acknowledge that WoS primarily indexes English-language articles and our search includes only English-language words.

The bibliographic information relating to all search results was imported into R for text analysis (R Core Team 2022). The search results were then limited to include only articles published between 1990 and 2023 and filtered to include only articles indexed with an abstract. Additionally, we created a subset of data from our selected journals (see Section 2.3).

The title and abstract text from each article were combined and tokenized by sentence using the Quanteda R package (Benoit et al. 2018), and the Quanteda keyword in context (KWIC) function was used alongside custom regular expressions to identify the presence of key phrases related to identify-first, person-first, and neurodiversity-related language (Data S1). Regular expressions are a sequence of numbers, characters, and/or symbols that can be used to identify patterns of text.

The number of publications which use each set of phrases was presented in graphs made using ggplot2 (Wickham 2016).

2.3 | Search Strategy and Article Selection Criteria

A systematic literature search was executed in the WoS Core Collection database, restricted to the selected journals of interest. Otherwise, the selection criteria were kept broad to allow a wide exploration of autism genetic articles. An initial search was limited to articles published (in press or print) in the last 5 years (2017–2021) to capture the current state of literature. The search was updated on February Tuesday 13, 2024. Search terms varied according to the type of journal to capture the genetic focus in autism journals, as well as the autism focus in genetic journals (Data S2). The identified articles were exported into Zotero, where titles and abstracts were screened by one independent reviewer (initial: H.K.K.; update: E.W.). All articles (including brief reports and proof of concept studies) that set out to investigate a genetic link to autism or autistic traits, with either autistic participants, a pre-existing dataset from autistic people, or animal or in vitro models, were included. Excluded articles were studies that did not investigate any genetic link to autism, or did not report new data and/or analysis. Articles that only made a genetic connection to autism post-hypothesis were also excluded. These criteria were specified in advance.

2.4 | The Development and Application of the Rating System

A novel rating system was developed to assess evidence of community engagement in the selected articles. This involved creating a signaling question for each of the three hallmarks of autism community engagement: (i) non-stigmatizing language, (ii) community-aligned priorities, and (iii) the use of participatory methods (Table 2). The rating system included five possible response options for each signaling question, ranging from "yes" or "probably yes," to "no" or "probably no," as well as an option for "no information." Next, exemplars for each hallmark and possible response were identified to help clarify the rating system and maximize consistency between raters. The rating system was applied to the full text of the total sample of screened and selected articles. This process involved reading each article in full, and then a targeted re-read while looking for evidence pertaining to each hallmark, and making a judgment about the appropriate rating based on the balance and consistency of evidence across the whole article. For instance, a single example of stigmatizing language would not be sufficient for a rating of "no" in response to the question: "is the language anti-stigmatizing?" Instead, the rating of "probably yes" or "probably no" was applied in cases where there was limited or inconsistent evidence. Articles with a mix of both stigmatizing and anti-stigmatizing

TABLE 2 | Rating system based on community-aligned indicators for determining level of engagement.

Signaling questions	Elaboration	Response options
1. Is there evidence of anti-stigmatizing language?	Answer "**yes**" if **identity-first** language is used, or if the **terms used** are **addressed** in the paper—for example, a **mix** of person-first and identity-first to cover all preferences of the autistic community. Other examples: (Bottema-Beutel et al. 2021): – Identity first language: "autistic," "person on the spectrum," "autistic person" – Neurodiversity – Higher likelihood/chance of developing – Non-autistic/neurotypical/comparison group – Description of specific needs and disabilities – Meltdown, stimming, specific description of behavior – Area of expertise, intense, passionate interest – Impact/effect rather than burden/suffer – Quality of life outcomes/discussions and mental health and wellbeing priority – Increasingly recognized/diagnosed Answer "**no**" if autism is consistently referred to with terms such as "**burden**," "**suffering**," "**epidemic**," "need of a **cure/recovery/optimal outcome**" (Gernsbacher et al. 2005; Kenny et al. 2016; Bottema-Beutel et al. 2021). Other examples: – Special interests – Special needs – Challenging/disruptive/problem behavior – High/low functioning; high/low severity – "At risk" for autism or "autism risk factors" (can be green if a single occurrence) – Autism symptoms—mild/high – Treatment – Healthy controls/normative sample – Deficit/disorder – Lack of empathy Answer "**no information**" if the paper does not refer or discuss autism at all with stigmatizing or anti-stigmatizing language. If there is a lack of evidence as described by the examples above. Answer "**probably no**" or "**probably yes**" if limited (see examples below) or inconsistent evidence is used throughout the paper. Predominant use of stigmatizing language but that includes occasional instances of non-stigmatizing language would receive a rating of "probably no." Conversely, the prevalence of non-stigmatizing language but some minor instances of stigmatizing language would warrant a "probably yes" rating. Other examples: – Only example of stigmatizing language is person-first language: "person with autism/ASD/ASC" (rating of "probably yes" if other examples of non-stigmatizing language is present, or rating of "probably no" if limited evidence) – Described as neuropsychiatric disorder (look at broader context, alone is not strong enough evidence for a rating of "no")	Y/PY/ PN/N/NI
2. Is there evidence of consideration of and/or alignment with community priorities?	Answer "**yes**" if the research priority is covered by the James Lind Alliance list of **top 10 priorities** (Cusack and Sterry 2016), or if autistic priorities are mentioned or discussed somehow, or explicitly stated (Pellicano et al. 2014a, 2014b; Nicolaidis et al. 2011) Examples: – Improve/reduce mental health problems and adapt mental health interventions for autistic needs – Language/communication skills development – Support/provide social care – Reduce anxiety – Best environment for achieving education/life/social skill outcome – Parents/family support/education for better understanding – Autism diagnostic criteria more relevant for adult population + appropriate diagnosis – Employers + workplace and autism – Better understanding of sensory processing – Service delivery	

(Continues)

TABLE 2 | (Continued)

Signaling questions	Elaboration	Response options
	Answer "**no**" if the priority does not align with the expressed priorities by the autistic community. Specific examples: – Findings can help design behavioral strategies for autism – Predictive biomarkers that can be used in prenatal screening – Drug targets for changing behavior of autistic children – Developing preventive strategies for autism – Outcomes focusing on curing autism – Outcomes focused solely on easing family members' "burden" Answer "**no information**" if priorities are not mentioned, covered, or explicitly stated in any regard. If there is lack of information about the priorities of the research. Examples: – Outcomes aimed at "improvements," but vague/broad descriptions used. Answer "**probably no**" or "**probably yes**" if it is unclear whether the priority mentioned is aligned with the autistic community or not. Examples: – "Probably yes" if priority is not stated, but the outcome/target aligns with one of the top 10 priorities (even if it is not explicitly recognized)	Y/PY/ PN/N/NI
3. Is there evidence of involvement of the autistic community through participatory methods?	Answer "**yes**" if paper uses **participatory methods** to include the autistic community such as **inclusive practices, emancipatory research, co-production, and community-based participatory research (CBPR)** (Nicolaidis et al. 2019; Fletcher-Watson et al. 2019) Examples: – **Inclusivity** (adapting the research environment, methodology and dissemination routes to permit the widest and most accessible engagement, or engagement from specific groups [e.g., nonspeaking autistic people and people with additional intellectual disabilities]) – **Emancipatory** (partnership, equal power and partnership, disseminate information out in the community) – **Co-production** (collaboration, e.g., co-created grant proposal with autistic co-applicants) – **CBPR**—advisory groups, autistic self-advocates as equal partners, for exampe, AASPIRE – **Consultation, citizen science, or leadership** Answer "**no**" if **participatory methods** have not been employed. – No evidence of any attempt made to include autism community views Answer "**no information**" if there is no information and a lack of evidence regarding how certain data were gathered or where consultancy came from. Examples: – Makes a claim of involvement, but not backed up by evidence – Worked with autistic people, but not stated how it influenced analysis Answer "**probably no**" or "**probably yes**" if it is unclear or include vague descriptions—such as: – Researchers believe they have involved the autistic community by qualitative data collection (e.g., interviews) – Researchers mention public engagement as a form of participatory research (vague)	Y/PY/ PN/N/NI

Abbreviations: N, no; NI, no information; PN, probably no; PY, probably yes; Y: yes.

language were assessed depending on the overall prevalence and context of the language used throughout the article.

Each article was coded against the rating system by one independent reviewer (initial: H.K.K.; update: E.W.) and 20% of the articles were double coded; disagreements were reconciled via discussion.

3 | Results

3.1 | Language Text Analysis

From our broad autism search, we identified 121,162 records. 118,177 records were published between 1990 and 2023, and of these 98,678 were indexed with an abstract.

Text analysis of the title and abstract text of these 98,678 records found that person-first language (e.g., person with autism) was the dominant phrasing used, although its usage appears to have dropped in the last year. Identity-first language (e.g., autistic person) appears less often but has been sharply rising in recent years (Figure 1A,B). Terms related to neurodiversity or phrasing such as "on the spectrum" appear to be less common in articles mentioning autism.

A subset of 4672 articles from the broad search were published in our selected autism journals, and 180 were published in our selected genetics journals. Although all articles in these subsets mentioned autism, not all referred to autism using identity- or person-first language and only those that matched our regular expressions are shown in Figure 1. Looking specifically at

the selected genetics journals, typically person-first language is used; however, identity-first language appeared in two publications from 2022 (Figure 1C). Within selected autism journals, person first language appears to be in decline, whereas identity-first language is on the rise (Figure 1D). The phrase "on the spectrum" and neurodiversity-related terms appear minimally.

3.2 | Screening Results

A total sample of 149 articles were included, comprising of 33 from genetics journals and 116 from autism journals (Figure 2; see Data S3 for a complete reference list). The genetic journal sample mostly consisted of studies investigating gene variants or mutations associated with autism and aimed to better understanding of the pathophysiology of certain mechanisms—in a tissue-specific manner or in relation to autism and health. Other studies within this category developed tools, methods and frameworks to analyze genetic information to optimize the scientific process and specifically applied them to autism case studies, cohorts and datasets. Similarly, the majority of the autism journal sample consisted of investigations into the functional relevance of genetic mechanisms involved in autism, either through identifying novel autism "risk" genes, examining polygenic risk scores, or comparing gene expression profiles of autistic populations and controls. A few studies examined the genetic mechanism underlying autism and its correlation to other co-occurring conditions such as sleep disturbance, ADHD, intellectual disability or gastrointestinal dysfunction, as well as epigenetic components.

3.3 | Rated Articles From the Genetics Journals

Figure 3 illustrates an overview of the rated articles from the genetics journals. Overall, minimal evidence of engagement was found in the 33 articles across the three areas captured, but particularly for the hallmarks of Considering Priorities and Reporting Engagement.

3.4 | Rated Articles From the Autism Journals

Figure 4 illustrates an overview of the rated articles from the autism journals. Here, there was greater evidence of engagement found in the 116 articles than in the genetics journals; however, this was still very rare.

3.5 | Proportional Evidence of Engagement

In terms of language use, 80% of articles used language scored as *stigmatizing* or *probably stigmatizing*, 14% used language that was *not* or was *probably not* stigmatizing, and in 6% there was not enough information to give the article a score. The priorities of 87% of the articles did *not* or *probably did not* align with autism community priorities, whereas only 13% were scored as *aligning* or *probably aligning* with research priorities. Only 1 article showed evidence that there was *probably* direct engagement with the autism community (Figure 5) and there were no definite examples of reported engagement.

To further elucidate these results, a selection of example articles, chosen to represent the range of ratings given, is presented in Table 3. These provide examples of the evidence found and used to generate the ratings.

4 | Discussion

This project aimed to investigate the exploratory hypothesis that minimal evidence of engagement with the autism community would be found in recently published literature on autism genetic research. This would be in line with autism community perspectives which cast doubt on the value of genetic research or express concerns over its potential uses (Natri et al. 2023). Overall, this project rarely found any evidence of engagement with the autism community within the three hallmarks identified and measured: non-stigmatizing language, alignment with community priorities, and the presence of direct engagement.

4.1 | Non-Stigmatizing Language

Text mining of article abstracts revealed person-first as the dominant choice of language to talk about autistic people and also revealed a recent sharp increase in the number of articles using identity-first language, suggesting a change is happening. Other language markers such as terminology associated with the neurodiversity paradigm were used very rarely indeed, and exclusively in articles published in the last few years. Similar trends can also be seen in the subset of autism and genetics journals that we investigated in greater detail. One reason for the lack of neurodiversity terminology might be that the neurodiversity movement is seen to be focused on autistic (and other neurodivergent) people who are articulate and intelligent and presumed therefore to have low support needs. Meanwhile, much genetic research focuses on autistic people with concurrent intellectual disability and obvious high support needs, because single-gene causes of autism are nearly always associated with concurrent intellectual disability (Zoghbi and Bear 2012). However, this is a misapprehension and the neurodiversity movement is concerned with the rights of all neurodivergent people, including the right to evidence-based medical treatment as needed (Chapman 2020), and includes many examples of autistic activists campaigning on behalf of those with less capacity for self-advocacy (e.g., National Autistic Taskforce, https://nationalautistictaskforce.org.uk/).

In autism and genetics journals, the majority of articles reviewed presented a consistent pattern of stigmatizing language. In most articles, the Diagnostic and Statistical Manual of Mental Disorders (DSM-5) (American Psychiatric Association 2013) was cited when authors characterized autism; however, the terminology employed varied greatly. Interestingly, some supplemented with more stigmatizing terms, whereas a scarce selection consciously chose non-stigmatizing terms. This illuminates the fact that the authors have a choice of language, even while drawing on authoritative sources such as the DSM-5. Examples of stigmatizing language include references to burden, suffering, risk, and severity. Instances of such language may arise from adherence to historical conventions and conformity to academic journal styles but also from the funding process. Researchers

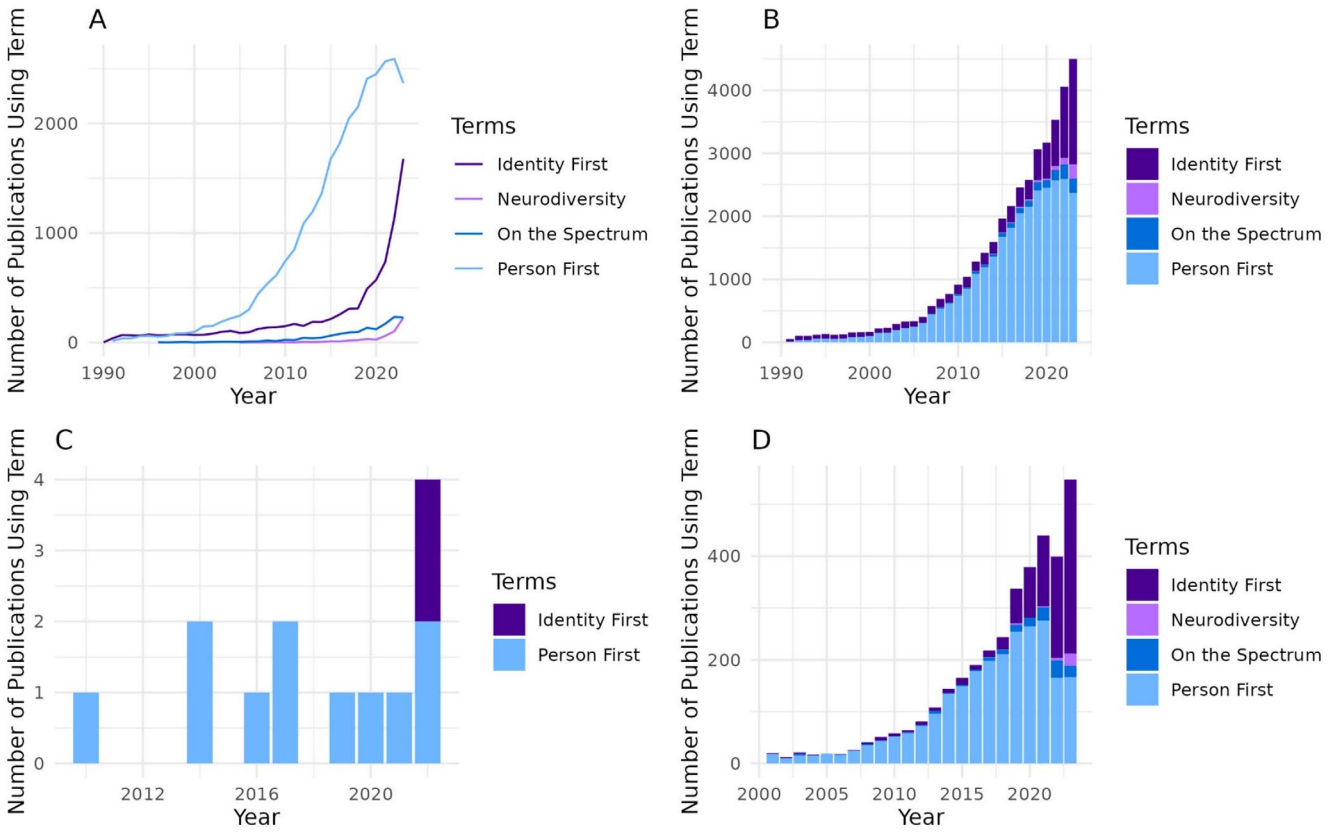

FIGURE 1 | Number of publications from broad WOS search using each set of phrases over time given as a line graph (A) and bar plot (B). Number of publications from selected genetics (C) and autism (D) journals over time.

may use the language of suffering and burden to highlight the importance of their research when in pursuit of funding, especially when competing for funding against teams studying, for example, the genetics of cancer or dementia. Funders and peer reviewers need to be cognizant of expectations around non-stigmatizing language in autism research, so that the use of respectful language does not result in a misapprehension of lesser need for research.

4.2 | Community-Aligned Priorities

We identified only a handful of examples of autism genetics papers that made reference to community priorities. These only achieved the level of probable indication, and centered on explicitly addressing the community (Besterman et al. 2020), targeting physical health outcomes (Riccio et al. 2018; Shindler et al. 2020) and aiming to deliver improvements in mental health (DiBlasi et al. 2020; Torske et al. 2020). No articles stated clearly and categorically that their goal was to deliver insights shaped by community priorities. The articles that received a "no" rating expressed research aims dedicated to providing molecular targets for therapeutic intervention or strategies, discovering candidate genes that contribute to autism likelihood and uncovering biological mechanisms related to the condition. These findings are consistent with existing evidence regarding a misalignment between the autistic community and autism research in the United Kingdom (Pellicano et al. 2014b). The autistic community want research to assist "with the day-to-day living with autism" (Pellicano et al. 2014b, 766), and genetics researchers should consider potential to deliver tangible and practical outcomes in their work, even if these may require additional translational research to be realized.

4.3 | Participatory Methods

We found no clear reporting of participatory methods across all articles and only one example with probable evidence of direct engagement. This example was a qualitative study investigating the perspectives of the autistic and autism community on autism biobanks. Lilley et al. (2024) specifically explored the experiences of autistic individuals, family members, and researchers who contributed to the Australian Autism Biobank, examining their motivations, concerns, and interpretations of autism genetic research. Although the study did not employ formal participatory approaches such as co-production, CBPR, or citizen science, it incorporated experiential expertise through the involvement of an autistic researcher in the data analysis process. The study engaged participants in discussions on autism genetics but did not explicitly adapt the research environment, methodology, or dissemination routes to enhance accessibility or promote direct involvement of autistic individuals in shaping the study. However, no reporting of direct engagement in the published articles does not necessarily amount to no involvement of the autistic community—it is possible that there was involvement of the autistic community, and that it merely was not reported. Recent publications have flagged the challenges of reporting community involvement and dedicated space to do so in journals remains rare (Tan et al. 2024; Fletcher-Watson 2024).

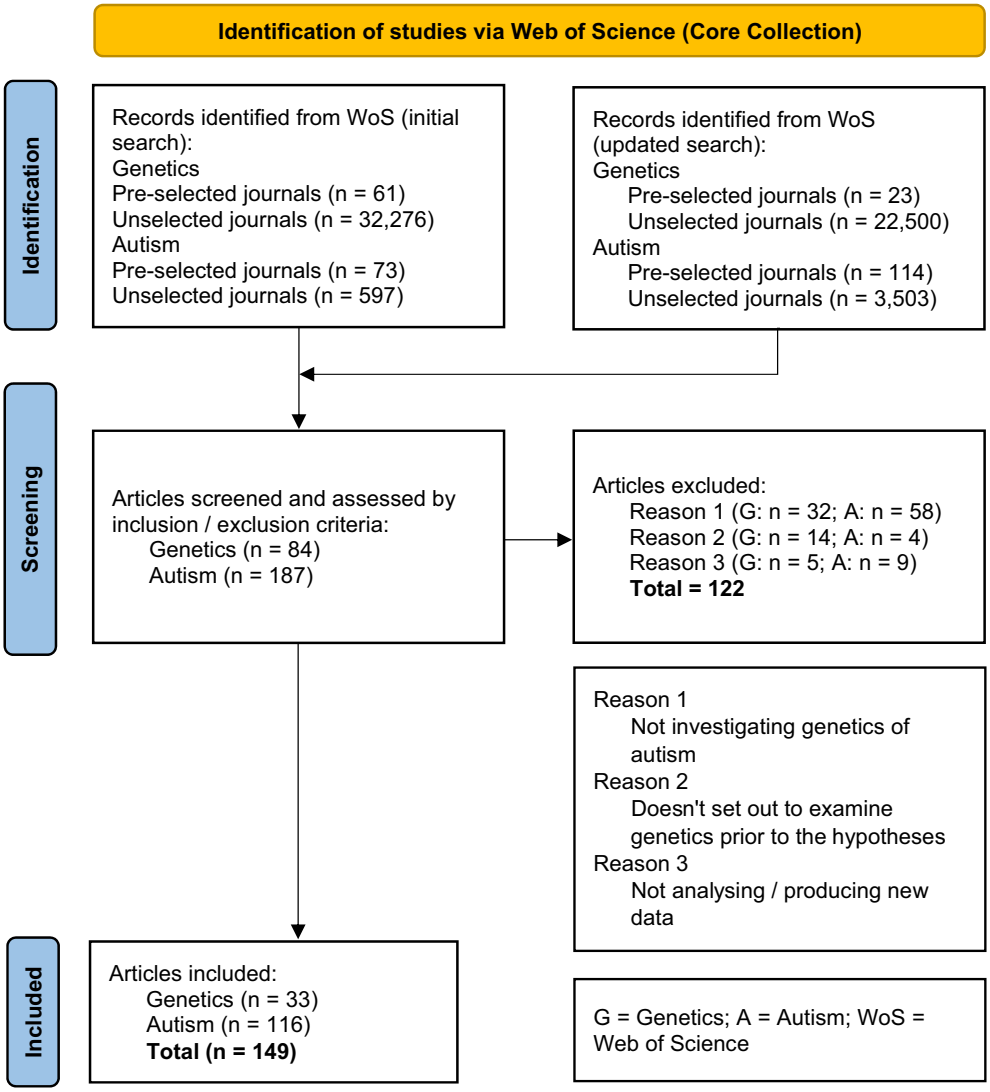

FIGURE 2 | Flow diagram of the article selection process (n = number of publications).

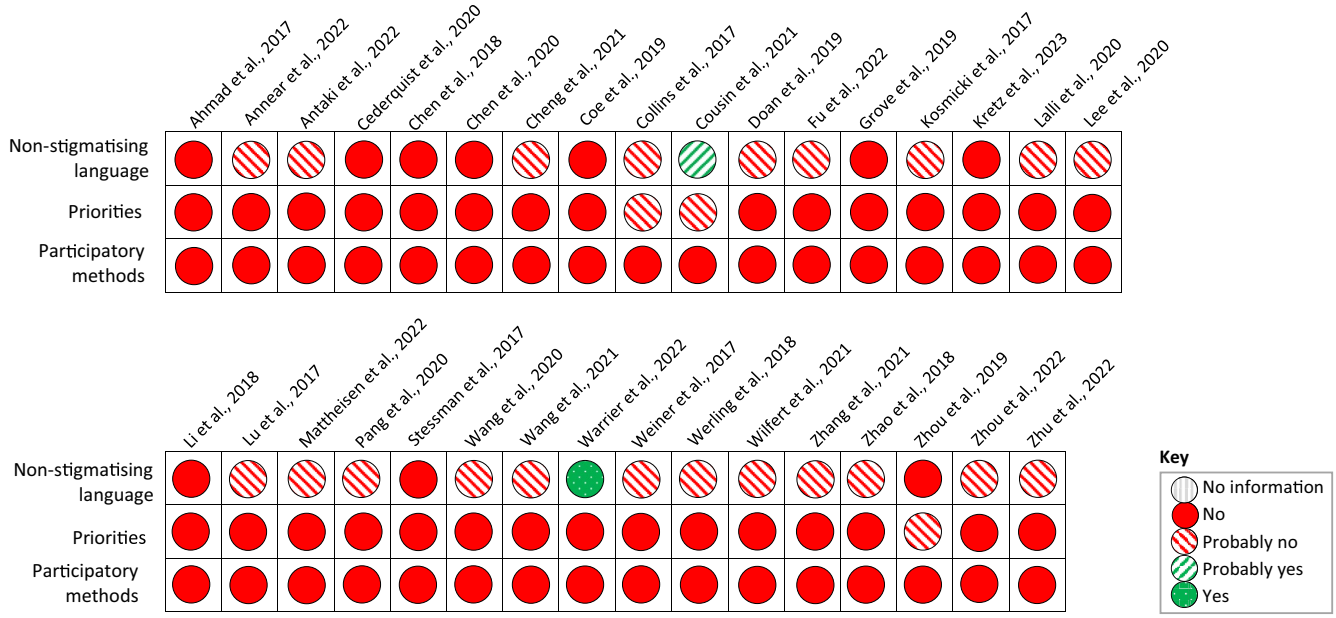

FIGURE 3 | Rated articles (n = 33) from the genetics journals.

FIGURE 4 | Rated articles ($n = 116$) from the autism journals.

Guidelines for participatory methods now abound but remain novel for the autism field (e.g., Nicolaidis et al. 2019). It may take some time for these practices to become established in genetics research which, by virtue of its highly technical nature, is challenging to make accessible to nonacademic stakeholders. In this context, the development of opportunities for autistic academics to lead work in this field is especially important. Many of the authors did thank families and participants in the "acknowledgements" of the articles, showing that researchers are not dismissive of their contributions, although there is little opportunity for community participation to hold more power.

4.4 | Limitations

The novel rating system used in this project was inspired by "risk of bias" tools, and devised for this project. Although we had good agreement on a double-coded sample, the results must be considered as preliminary until the robustness of the rating scheme can be established further. Due to the practical constraints, we examined evidence of engagement in influential papers mainly determined by IF and SJR metrics, and consequently excluded other relevant articles. However we don't expect that other genetics papers published in less highly ranked journals would have differed systematically from our sample in their evidence of engagement. Where we have sought to identify community engagement, our results are limited by reporting of any engagement in published research articles, that is, if research included some aspect of community engagement, but this was not stated in subsequent publications. We are unsure to what scale, if at all, this may impact our findings, and we would note that failure to report community engagement in the research process in publications if a problem in itself. The results are also limited by inclusion of only articles printed in English.

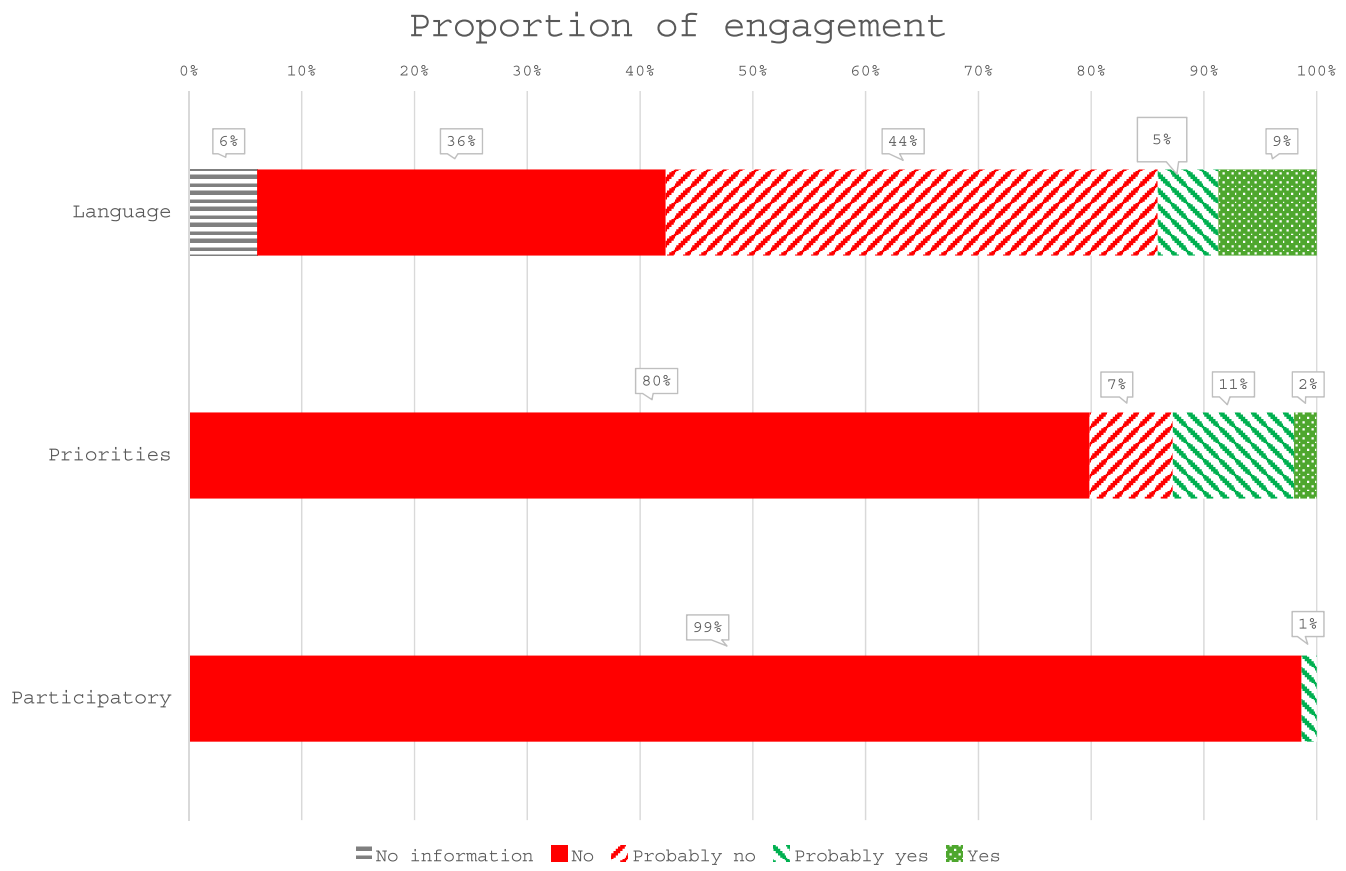

FIGURE 5 | Proportion of engagement graph, showing percentage of engagement in the total sample of articles within the three domains.

Finally, this project itself was not co-produced with the autistic community, though it was based on co-produced work and autistic writing to set the hallmarks. There might be other hallmarks of engagement that autistic people would look for in the literature.

4.5 | Future Directions

The overarching pattern observed in these results signifies a lack of engagement with the autism community in autism genetic research, which can have repercussions for autism genetic researchers. These findings can help explain negative responses of the autistic community to some genetic research (Sanderson 2021; Natri 2021). If this lack of engagement is not addressed, it may result in study disruption, loss of trust between researchers and the intended beneficiaries, and ultimately bias results. For instance, if autistic adults who are consenting for themselves decline to participate in genetic research, the participant sample might get skewed toward autistic children and adults with an intellectual disability. This would be a serious issue for discovery science, because intellectually able autistic people are diagnosed later in life (Atherton et al. 2021), and autistic people with an intellectual disability are more likely to have a monogenic cause (Jeste and Geschwind 2016). We recommend that genetic autism researchers immediately start to address the stigmatizing language and terms used, and start to develop greater engagement and inclusion of the autism community in their work. This is especially important when investing large sums in long-term research programs: Involvement can aid in defining more relevant research questions for a population, thus avoiding unnecessary research (Beresford 2002; Chalmers and Glasziou 2009). Autistic scientists with knowledge of genetic research methods and terminology should be central to this effort. Identifying barriers to collaborative working, addressing concerns about future misuse of biobank data (Lilley et al. 2024), and setting specific priorities for the field would all be good places to start. Optimally, there would be a flagship co-produced project that would set a standard and model for the field. To support this work, more in-depth analysis of the variability in engagement in the field (e.g., by study country or specific population or method, such as polygenic architecture versus genetic syndrome) and qualitative data collection with genetics researchers exploring barriers to participatory working would add value.

5 | Conclusions

In conclusion, this study found minimal evidence of engagement with the autistic community in current autism genetic research, as measured by automated text analysis and a novel rating system. The raised concerns by the autism community are not unwarranted. Although there is some evidence of an increase in the use of non-stigmatizing language, stigmatizing language is still highly prevalent in the published literature. We strongly encourage autism researchers to avoid using stigmatizing language as a prerequisite to building trust. A greater involvement and engagement with that community could lead to more successful delivery of autism genetic research and greater translational potential. Conversely, a lack of engagement can lead to

TABLE 3 | Summary table of evidence of engagement.

Author, publication year	J	Language	R	Priorities	R	Participatory methods	R
Chen et al. 2020	G	"Individuals with ASD (p.1)," "... characterized by limited social communication... (p.1)," "contribute to ASD susceptibility (p.7)," "developing potential therapeutic strategies to address these neuropsychiatric disorders, including ASD (p.12)"	N	Develop therapeutic strategies	N	No evidence	N
Lalli et al. 2020	G	"ASD-associated genes (p.5)," "ASD-causing genes (p.5)"	PN	Aimed to assess potentially convergent mechanisms in ASD and link molecular pathways to clinical phenotypes	N	No evidence	N
Zhao et al. 2018	G	"Contribute to ASD risk (p.1)," "develop therapeutics for ASDs (p.1)," "ASD individuals (p.6)," "autistic individuals (p.6)"	PN	Develop therapeutics for ASDs	N	No evidence	N
Arnett et al. 2019	A	"Atypical social communication (p.1)," "autism patients (p.2)," "genetic subtypes of ASD are associated with unique behavioral, medical and cognitive phenotypes (p.2)," "ASD symptoms (p.2)," "children with ASD (p.2)," "knowledge about how early development predicts later functioning promotes proactive, targeted interventions, as well as opportunities for psychoeducation and social emotional support (p.2)," "we expect that the exploratory analyses conducted in this study will provide preliminary evidence on which to base future work, including development of guidelines for clinicians and families seeking to support and better manage affected individuals across the lifespan (p.2)"	PY	Identify developmental milestones that predict cognitive and adaptive outcomes for five of the most common ASD genotypes. The research aims to support personalized treatment planning and long-term care strategies. The findings are intended to provide families and clinicians with valuable insights for improving intervention timing, educational planning, and overall support for individuals with autism	PY	No evidence	N
DiBlasi et al. 2020	A	"Individuals with ASD compared with non-ASD populations (p.1)," "social communication difficulties (p.1)," "individuals with ASD (p.5)"	PN	Understand ASD biological risk and better identify those at risk for suicidal behavior, develop targeted personalized interventions to prevent suicide risk for suicide	PY	No evidence	N
Lewis et al. 2018	A	"Neurotypical sample (p.1)," "ASD characterized... socio-cognitive deficits (p.1)," "face memory deficits (p.1)," "children with ASD diagnosis (p.2)"	N	Understand autism-like phenotype to investigate deficits in face memory ability	N	No evidence	N

(Continues)

TABLE 3 | (Continued)

Author, publication year	J	Language	R	Priorities	R	Participatory methods	R
Massrali et al. 2019	A	"(Autism characterized) by social-communication difficulties, unusually restrictive, repetitive behavior and narrow interests and sensory difficulties (p.1)," "more male autistic individuals (p.11)," "autistic individuals (p.11)," "neurotypical (p.11)"	Y	To understand autism and autistic trait overlap by comparison of DNA methylation at birth and scores capturing autistic traits	PN	No evidence	N
Schendel et al. 2022	A	"ASD is marked by deficits in social communication and restricted, repetitive behaviors (p.2)," "risk for ASD (p.5)," "autistic females or autistic persons with ID tended to have lower ASD PRS means than autistic males or autistic persons without ID (p.5)," "clinical risk prediction efforts for ASD... as complementary measures of family-based autism risk (p.10)"	N	Efforts to enhance clinical risk prediction for ASD by looking at interrelations between psychiatric family history and individual ASD genetic liability	N	No evidence	N
Shindler et al. 2020	A	"The genetics of autism is complex with a broad range of biomarkers correlating with the severity of symptoms but no single biomarker able to be used for diagnosis (p.2)," "patients with autism (p.2)," "to identify the prevalence of specific SNPs within the autism community (p.2)," "ASD participants (p.6)," "the participants with autism and the neurotypical GI dysfunction participants (p.7)," "the autistic group (p.4)," "smaller autistic sample size (p.8)," "in autistic populations (p.8)"	PY	Aims to identify genetic biomarkers associated with gastrointestinal dysfunction in autistic individuals to enhance understanding of genetic risk factors and contribute to the development of diagnostic tools. It focuses on analyzing specific single nucleotide polymorphisms (SNPs) related to GI symptoms with the goal of improving symptom management and informing potential interventions	PY	No evidence	N
Lilley et al. 2024	A	"Autistic and autism communities (p.1)," "autistic and non-autistic children (p.2)," "autistic participants (p.2)," "The search to prevent or cure autism, once an explicit aim of some autism research, is now considered by many in the research and advocacy communities to be undesirable, and some advocacy organizations have sharply distanced themselves from it (p. 2)," "the narratives of biobank participation would offer a unique contribution to understanding attitudes toward autism science and the "contextual bioethics (p. 2)"	Y	Investigates the perspectives of autistic individuals, their families, and researchers on contributing to an autism biobank. The study aims to understand participants' motivations, concerns, and experiences with genetic research and biobanking. It explores themes such as the search for causes of autism, ethical considerations surrounding biobanks, and community trust in scientific research	Y	Study engaged autistic participants and family members in interviews to gather their views and reflections on biobank participation. Although there is evidence of meaningful engagement, it does not extend to full co-production of research. The researchers took efforts to incorporate autistic voices, such as including an autistic researcher in the analysis phase (p.5), but the study primarily remains researcher-led	PY

Abbreviations: A, autism; G, genetic; J, journal; N, no; NI, no information; PN, probably no; PY, probably yes; R, rating; Y, yes.

study disruption, mistrust in the scientific process, wasted investment, and unfruitful research outcomes.

Author Contributions

H.K.K.: Data curation; Formal analysis; Investigation; Project administration; Validation; Visualisation; Writing – original draft. S.F.-W.: Conceptualisation; Supervision; Writing – review and editing. E.W.: Investigation; Validation; Visualisation; Writing – review and editing.

Ethics Statement

The authors have nothing to report.

Conflicts of Interest

The authors declare no conflicts of interest.

Data Availability Statement

The dataset and materials supporting the conclusions of this article are included within the article and its Supporting Information. Analysis code for text mining is available under a Creative Commons by Attribution 4.0 license: https://github.com/emma-wilson/kaljusto-2025-text-mining.

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Supporting Information

Additional supporting information can be found online in the Supporting Information section.