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A multidimensional exploration of gender nonconformity and gender dysphoria

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Abstract

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Gender nonconformity refers to a condition where an individual's gender expression or identity does not align with societal expectations associated with their sex assigned at birth. Gender dysphoria is characterized by distress experienced as a function of a significant mismatch between a person's experienced or expressed gender and the sex assigned at birth. These conditions are influenced by biological, psychological, and sociocultural factors, with many unknowns regarding their developmental aspects. This thesis aimed to explore these two phenomena using a multidisciplinary approach from various perspectives. Data from two cohort studies, the Swedish Environmental Longitudinal Mother and Child Asthma and Allergy study and the Swedish Gender Dysphoria Study, as well as interviews with individuals experiencing gender dysphoria, were utilized.

Biological correlates of gender nonconformity were examined through the relationship between prenatal phthalate exposure and gender-specific play behavior, with suggestive associations observed. Associations between gender nonconforming play behavior and behavioral difficulties were also identified in children. Childhood gender nonconformity was linked to gender dysphoria and decreased mental well-being in individuals with gender dysphoria. In terms of the connection between gender diversity and autism, gender nonconforming play behavior was associated with autistic traits in girls, and individuals with gender dysphoria exhibited a higher prevalence of autism spectrum disorder and elevated autistic traits compared to cisgender individuals. Finally, gender identity formation was characterized as a dynamic, non-linear, and individualized process among those experiencing gender dysphoria, articulated through the variety of terms used by these individuals to label their gender identity. In conclusion, this thesis contributes important knowledge to understanding gender nonconformity and gender dysphoria.

Keywords: gender identity, gender nonconformity, gender dysphoria, endocrine disrupting chemicals, mental health, autism

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This thesis was written by the author without content generation using text generative artificial intelligence (AI) tools. Where AI was used, it was in line with the Department of Organismal Biology's policy, such as for grammar and style refinement, translation, or coding assistance, with full oversight and verification by the author.

List of Papers

This thesis is based on the following papers, which are referred to in the text by their Roman numerals.

I. **Özel, F.**, Stratmann, M., Lindh, C., Gennings, C., Bornehag, C. G., & Rüegg, J. (2023). Prenatal exposure to phthalates and gender-specific play behavior at seven years of age in the SELMA study. *Environment International, 178*, 108029.

II. **Özel, F.***, Stratmann, M.*, Papadopoulos, F. C., Rüegg, J., & Bornehag, C. G. (2024). Gender-specific play behavior in relation to autistic traits and behavioral difficulties at the age of seven in the SELMA study. *PLOS One, 19*(8), e0308605.
**contributed equally*

III. **Özel, F.**, White, R. A., Johansson, M., Rüegg, J., & Papadopoulos, F. C. Interplay between childhood gender nonconformity and later life gender dysphoria and mental health. *Manuscript under review.*

IV. **Özel, F.**, White, R. A., Clark, K. D., Indremo, M., Zejlon, I., Rüegg, J., & Papadopoulos, F. C. Associations between autism, gender dysphoria, and gender incongruence: insights from the Swedish Gender Dysphoria Study (SKDS). *Manuscript under review.*

V. **Özel, F.**, Griffin, G., & Rüegg, J. The Words to Say It: gender dysphoria, gender identifications, trans and people aged 15-35 in Sweden. *Manuscript under review.*

Additional Papers

The following papers were published during my doctoral education but are not included in this thesis:

Stratmann, M., **Özel, F**., Marinopoulou, M., Lindh, C., Kiviranta, H., Gennings, C., & Bornehag, C. G. (2024). Prenatal exposure to endocrine disrupting chemicals and the association with behavioural difficulties in 7-year-old children in the SELMA study. *Journal of Exposure Science & Environmental Epidemiology*, 10.1038/s41370-024-00739-x. Advance online publication.

Özel, F., Di Criscio, M., Lupu, D. I., Sarkisyan, D., Hlady, R. A., Robertson, K. D., Bakalkin, G., Liu, Y., Biernacka, J. M., Karpyak, V. M., Ekström, T. J., & Rüegg, J. (2024). DNA methylation at DLGAP2 and risk for relapse in alcohol dependence during acamprosate treatment. *Drug and Alcohol Dependence, 256*, 111116.

Özel, F., Indremo, M., Karamanis, G., Elofsson, U., Beckman, U., Fazekas, A., Frisén, L., Isaksson, M., Sandström, L., Thelin, N., Tivesten, Å., Wahlberg, J., Skalkidou, A., Bodlund, O., & Papadopoulos, F. C. (2023). Exploring gender dysphoria and related outcomes in a prospective cohort study: protocol for the Swedish Gender Dysphoria Study (SKDS). *BMJ Open, 13*(4), e066571.

Özel, F., & Rüegg, J. (2023). Exposure to endocrine-disrupting chemicals and implications for neurodevelopment. *Developmental Medicine and Child Neurology, 65*(8), 1005–1011.

Cediel-Ulloa, A., Lupu, D. L., Johansson, Y., Hinojosa, M., **Özel, F.**, & Rüegg, J. (2022). Impact of endocrine disrupting chemicals on neurodevelopment: the need for better testing strategies for endocrine disruption-induced developmental neurotoxicity. *Expert Review of Endocrinology & Metabolism, 17*(2), 131–141.

Contents

Abbreviations

AFAB	Assigned female at birth
AMAB	Assigned male at birth
APA	American Psychiatric Association
AQ	Autism Spectrum Quotient
ASD	Autism spectrum disorder
BPA	Bisphenol A
CAH	Congenital adrenal hyperplasia
CAIS	Complete androgen insensitivity syndrome
CI	Confidence interval
DEHP	Di-2-ethylhexyl phthalate
DiNP	Di-isononyl phthalate
DSD	Differences in sex development
DSM	Diagnostic and Statistical Manual of Mental Disorders
EDC	Endocrine disrupting chemical
EMB	Extreme male brain
ICD	International Classification of Diseases
MBP	Mono-butyl phthalate
MBzP	Mono-benzyl phthalate
MCiNP	Mono-carboxy-isononyl phthalate
MCiOP	Mono-carboxy-isooctyl phthalate
MCMHP	Mono-2-carboxymethylhexyl phthalate
MD	Mean difference
MECPP	Mono-2-ethyl-5-carboxypentyl phthalate
MEHHP	Mono-2-ethyl-5-hydroxyhexyl phthalate
MEHP	Mono-2-ethylhexyl phthalate
MEOHP	Mono-2-ethyl-5-oxohexyl phthalate
MEP	Mono-ethyl phthalate
MHiDP	Mono-hydroxy-isodecyl phthalate
MHiNP	Mono-hydroxy-isononyl phthalate
MOiNCH	2,4-methyl-7-oxyooctyl-oxycarbonyl-cyclohexane carboxylic acid
MOiNP	Mono-oxo-isononyl phthalate
OR	Odds ratio

PSAI	Preschool Activities Inventory
RAADS-14	Ritvo Autism and Asperger Diagnostic Scale-14
RCGI	Recalled Childhood Gender Identity/Gender Role Questionnaire
RHV	Repeated holdout validation
SDQ	Strengths and Difficulties Questionnaire
SELMA	Swedish Environmental Longitudinal Mother and Child Asthma and Allergy (study)
SKDS	Swedish Gender Dysphoria Study (Svenska Könsdysforistudien)
SRS	Social Responsiveness Scale
TCS	Transgender Congruence Scale
TGD	Transgender and gender diverse
WHO	World Health Organization
WQS	Weighted quantile sum regression

Introduction

In recent years, gender diversity has become a significant and widely discussed topic in both academic and public discourse. Academically, there has been a prominent interest regarding gender diversity in fields such as psychiatry, psychology, and other social science disciplines. This growing interest has had a profound impact on medical and social perspectives on gender identity.

Academic research has played a fundamental role in shaping the narratives on gender diversity by providing insights into its biological, psychological, and sociocultural dimensions. Increasingly, there has been an emphasis on the multifaceted nature of the phenomenon with more focus on the intersectionality of different dimensions. Despite substantial progress in this field, particularly over the past two decades, our knowledge of gender diversity remains rather limited, necessitating multi- and interdisciplinary perspectives.

In line with the accumulating research findings and existing knowledge gaps with regard to gender diversity, this current work adopts a multidisciplinary approach and explores gender nonconformity and gender dysphoria from multiple angles. In the following sections, I provide relevant background information including on the terminology used in the field, epidemiological findings, developmental perspectives, and current knowledge about coexisting mental health conditions, and outline the research aims. I then describe the methodology and summarize individual study results, followed by a general discussion.

Terminology

Changes in the understanding of gender and gender identities have led to shifts in vocabulary in recent years. Still, certain basic terms, including sex, sex assigned at birth, gender, and gender identity are sometimes used interchangeably even when this is inappropriate. Moreover, terms related to gender diversity are often used in academic texts without clear definitions. This can result in the same term having various meanings and potentially misleading readers.

Therefore, I begin by defining the key terms I use and explaining the context in which I use them.

Sex refers to biological characteristics, and sex assigned at birth is used to define a person's status of being female, male, or intersex based on their physical features, most commonly on the appearance of their external genitalia after birth (Coleman et al., 2022). Assigned female at birth (AFAB) and assigned male at birth (AMAB) are used in this thesis to refer to sex assigned at birth. Gender, on the other hand, refers to the socially constructed characteristics of women, men, girls, and boys according to the definition of the World Health Organization (WHO, 2025), and is connected to norms, behaviors, and roles. Gender identity is defined by the American Psychological Association (2015) as a person's deeply felt, internal sense of being a girl, a woman, or female; a boy, a man, or male; or an alternative gender.

One's sex assigned at birth and gender identity do not necessarily align with each other. The term cisgender describes people whose sex assigned at birth and gender identity align (American Psychological Association, 2015). Transgender and trans are umbrella terms used to refer to people whose gender identity, and in some cases gender expression, does not correspond with the expectations typically associated with their sex assigned at birth (Coleman et al., 2022). Gender diverse is a term used to describe individuals with gender identities that are different from the social and cultural expectations attributed to their sex assigned at birth (Coleman et al., 2022). This term can be used more broadly to encompass all individuals with different gender identities who do not identify as cisgender. In this thesis, I do not use the terms transgender and gender diverse synonymously.

Vocabularies used to define gender and gender(ed) identities have proliferated drastically since the late 1990s (Thorne et al., 2019), and non-binary perspectives on gender have been articulated more broadly. The term non-binary refers to a heterogenous group of people whose gender identity exists between or beyond the categories of female and male, people who may experience being a woman and a man at different times, and those who do not identify with any gender, among others (Matsuno & Budge, 2017). Along with the term non-binary, various terms such as genderqueer, genderfluid, and gender expansive have also emerged to describe divergent gender identities. The definitions of these are not provided here, first because this thesis is not specifically focused on them, and second because there is no consensus regarding their use in academic texts. Scholars use this vocabulary in various contexts to indicate different constructs. This not only weakens the terms' descriptive ability but also leads to particular assumptions about the gender identities of people who identify with these terms. In fact, empirical evidence on how gender diverse people perceive these terms and in which terms they describe their gender identities are relatively scarce.

Gender nonconformity is one of the main concepts this thesis focuses on. Gender nonconformity describes a state where a person's gender expression or gender identity do not conform to the gender norms linked with their sex assigned at birth (American Psychological Association, 2015). Specifically, gender nonconformity can cover various types of expressions, behaviors, and roles, and nonconforming traits may or may not reflect particular gender identities. Gender nonconformity is sometimes used more broadly to comprise people whose gender identity differs from their sex assigned at birth; however, in this thesis, I use gender nonconformity only to refer to gender expressions, behaviors, and roles that do not align with gender norms and societal gender expectations.

In contrast to the terms mentioned above, gender dysphoria and gender incongruence concern clinical diagnoses. Gender dysphoria is defined as "a marked incongruence between one's experienced/expressed gender and assigned gender, of at least six months' duration" according to the Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-5; American Psychiatric Association [APA], 2013). This diagnostic category also requires the presence of clinically significant distress. DSM-5 has two sets of diagnostic criteria, one for children and one for adolescents and adults. Before DSM-5 came into use in 2013, the same condition was called gender identity disorder with some differences in the diagnostic criteria (APA, 2000). Gender incongruence, on the other hand, is a clinical diagnosis according to the 11th revision of International Classification of Diseases (ICD-11) by the WHO (2019). In ICD-11, the previous diagnostic category of gender identity disorder was replaced with gender incongruence which is defined as "a marked and persistent incongruence between an individual's experienced gender and the assigned sex" (WHO, 2019). ICD-11 similarly distinguishes the criteria for children and adolescents or adults, and the discussion of gender incongruence is situated in a chapter about conditions related to sexual health, whereas previous diagnoses were in the chapter on mental and behavioral disorders. Unlike DSM-5, the criterion for feeling distressed was removed in ICD-11. This allows to cover individuals who experience incongruence between their gender identity and sex assigned at birth but do not feel significant discomfort.

The use of specific terminologies may show some variations in different contexts. Although the term gender dysphoria in this thesis refers to a clinical diagnosis, it has historically been used to describe the discomfort caused by a misalignment between experienced and assigned gender, even for people who do not meet the diagnostic criteria (Byne et al., 2018). Additionally, not all transgender and gender diverse (TGD) people experience such discomfort (Byne et al., 2018). In this thesis, the term gender dysphoria is used to refer to people who experience distress due to a discrepancy between their gender identity and sex assigned at birth; they may or may not have a clinical diagnosis, depending on the context. The term TGD is used to indicate a broader group of individuals, with or without distress. The terms gender diversity and gender variance are used interchangeably. Lastly, other terms used in the field may occur in the following sections, where I keep the original terminology from the referenced articles.

Epidemiology

Studies investigating population estimates of TGD people have employed different information sources, including healthcare-based and population-based data, as well as various definitions regarding gender diversity, gender nonconformity, and gender dysphoria. Although it is challenging to draw definitive conclusions due to discrepancies across studies, two literature reviews on this topic have provided important findings about population estimates concerning gender diversity. Based on data collected in 17 countries between 1968 and 2018, Goodman et al. (2019) concluded that the proportion of TGD people in the general population ranged from 0.1% to 2%. In a systematic review focusing on studies conducted between 2009 and 2019, Zhang et al. (2020) have shown that the proportion of TGD individuals based on healthcare data ranged between 17 and 33 per 100,000 persons. The proportion of having transgender identity was 0.3-0.5% among adults and 1.2-2.7% among children and adolescents in survey-based data (Zhang et al., 2020). The same study also reported the proportion of gender diversity, assumed as a broader concept than transgender, which was 0.5-4.5% among adults and 1.5-8.4% among children and adolescents (Zhang et al., 2020). Despite increasing knowledge about population estimates of gender diversity and gender dysphoria, current evidence mostly refers to western Europe and north America, which limits its generalizability.

Regarding the population estimates in Sweden, 0.06% of the Swedish population had a gender dysphoria diagnosis in 2018 (Socialstyrelsen, 2020). Starting from 2021, between 800 and 1,200 individuals were newly diagnosed with gender dysphoria each year, as reported recently by the National Board of Health and Welfare in Sweden (Socialstyrelsen, 2025). In a population-based study conducted among adults from Stockholm County, 2.3% of the participants reported feeling like someone of a different sex, while 2.8%

reported a desire to live as or be treated as a person of another sex (Åhs et al., 2018).

An increase in the number of clinical referrals for gender dysphoria has been documented in several countries in recent years (Goh et al., 2024; Jarvis et al., 2025; Kaltiala et al., 2020; Nyquist et al., 2025; Wiepjes et al., 2018). A more significant increase has been observed for individuals AFAB compared to AMAB (Aitken et al., 2015; Jarvis et al., 2025; Kaltiala et al., 2023; Nyquist et al., 2025; Zhang et al., 2021). This contradicts the earlier findings that individuals AMAB were referred for gender dysphoria more frequently than AFABs (Steensma et al., 2018). Trends in the rise of gender dysphoria diagnoses have also been demonstrated in Sweden (Claesdotter-Knutsson et al., 2024; Socialstyrelsen, 2025). Between 2004 and 2015 in Sweden, Indremo et al. (2021) reported a significant increase in gender dysphoria diagnoses among individuals AFAB, with a comparatively smaller but still notable rise observed among those AMAB.

The reasons behind the recent demographic changes in gender dysphoria diagnoses remain unknown. Various factors have been suggested, such as greater awareness of the phenomenon, less stigma associated with seeking treatment, and changes in diagnostic criteria, among others (Claesdotter-Knutsson et al., 2024; Indremo et al., 2021). Regardless of the underlying reasons, these changes and increased salience highlight the importance of improving our understanding of gender dysphoria.

Developmental perspectives

Since the initial works focusing on differences between genders, numerous studies have investigated the development of gender, gender identity, gender nonconformity, and gender dysphoria from several perspectives, including biological, psychological, and sociocultural ones. In fact, all these different aspects play a role in gender identity formation rather than a sole developmental origin (de Vries et al., 2014). At present, gender identity is viewed as a multifaceted and dynamic construct influenced by a variety of factors (McGuire & Morrow, 2020).

Biological influences

Biological factors are considered to contribute to gender nonconformity and gender dysphoria to varying extents. Many researchers have focused on the potential role of genetics; some degree of heritability of gender nonconformity, gender dysphoria, or gender diversity has been supported by previous studies (Diamond, 2013; Iervolino et al., 2005; Knafo et al., 2005; Sasaki et al., 2016). Associations for genetic polymorphisms in sex hormone receptors have been shown among TGD individuals (Fernández et al., 2014; Fernández et al., 2018; Foreman et al., 2019; Henningsson et al., 2005). Additionally, differences in epigenetic modifications between transgender and cisgender people have been proposed (Ramirez et al., 2021). Still, evidence on the role of genetic and epigenetic factors is limited and inconclusive.

Another focus area of the neurobiological mechanisms underlying gender identity, gender nonconformity, and gender dysphoria is sex hormones. Based on early animal studies showing a role for sex hormones in the sexual differentiation of the brain (Arnold, 2009), researchers examined medical conditions where the fetus was exposed to high levels of sex hormones during gestation. Gender roles and expressions have been investigated among children with congenital adrenal hyperplasia (CAH) and complete androgen insensitivity syndrome (CAIS). These are genetic conditions that cause either high levels of testosterone or insensitivity to androgens, respectively (Delli Paoli et al., 2023; Engberg et al., 2025). Increased levels of masculine behavior, decreased levels of feminine behavior, and higher proportions of gender diversity have been reported among individuals AFAB with CAH in comparison to those AFAB without CAH (Kung et al., 2024; Meyer-Bahlburg et al., 2006; Nordenström et al., 2002; Pasterski et al., 2011; Pasterski et al., 2015; Servin et al., 2003). Among individuals with an XY karyotype and CAIS, heightened feminine behavior and identification have been reported (Hines et al., 2003; Jürgensen et al., 2007). However, it is crucial to acknowledge that these studies have been conducted with participants with differences in sex development (DSD). Studies examining the effects of testosterone within physiological levels yielded less consistent results (Auyeung et al., 2009; Hines et al., 2002; Knickmeyer et al., 2005; Lamminmäki et al., 2012; van de Beek et al., 2009). Besides, current evidence has mostly concentrated on gender roles and expressions, and findings on gender identity and gender dysphoria are rather limited.

Based on the evidence that sex hormones may play a role in gender identity development, a potential link between prenatal exposure to endocrine disrupting chemicals (EDCs) and gender dysphoria has been claimed (Bejerot et al., 2011; Hood, 2005). The WHO (2002) defines an EDC as “an exogenous substance or mixture that alters function(s) of the endocrine system and consequently causes adverse health effects in an intact organism, or its progeny, or (sub)populations”. EDCs can interfere with hormonal functions at different levels; for instance, they can act on endocrine cells or hormone receptors, change hormone metabolism or transport, or affect intracellular hormone-related regulations (La Merrill et al., 2020). EDCs represent a large group of chemicals, including organochlorine compounds, halogenated aromatic hydrocarbons, brominated flame retardants, per- and polyfluoroalkyl substances, alkylphenols, phthalates, and bisphenol A (BPA) and its analogues (Metcalfe et al., 2022). EDCs are used, among others, to produce consumer goods, food packaging materials, personal care products, and cosmetics. Humans are constantly exposed to EDCs through these sources (La Merrill et al., 2020). Experimental and epidemiological evidence has clearly shown that prenatal exposure to EDCs is associated with various adverse health outcomes, such as obesity, neurodevelopmental issues, and cancer (Kahn et al., 2020; Metcalfe et al., 2022; Modica et al., 2023).

Even though a potential link between EDCs and gender dysphoria has been repeatedly mentioned in the literature, scientific evidence supporting this link is lacking. Two studies investigated whether prenatal exposure to diethylstilbestrol or anticonvulsants was associated with diverse gender identities later in life (Dessens et al., 1999; Troisi et al., 2020). However, the very small number of gender diverse people in these studies makes it difficult to draw meaningful conclusions. Another recent study examined the concentrations of two insecticides, thiamethoxam and fipronil, and BPA among people diagnosed with gender dysphoria and cisgender controls (Üstay et al., 2024). They found that AFAB diagnosed with gender dysphoria had higher levels of thiamethoxam in comparison to cisgender AFABs, but not BPA or fipronil. However, this study had a cross-sectional design and did not provide information on prenatal exposure, which has been suggested as the critical developmental timespan for the potential connections between EDCs and gender dysphoria.

Within this field of research, associations between prenatal exposure to EDCs and gender-specific roles and behaviors have been investigated more extensively than potential links with gender dysphoria. In two studies, dioxins and polychlorinated biphenyls have been shown to be related to alterations in gender-specific play behavior scores measured by the Preschool Activities Inventory (PSAI) (Vreugdenhil et al., 2002; Winneke et al., 2014). Phthalates have been a specific focus of interest because of their well-known anti-androgenic properties; associations between prenatal exposure to phthalates and gender roles and behaviors have been indicated (Evans et al., 2021; Liu et al., 2023; Percy et al., 2016; Swan et al., 2010). However, the findings are limited and inconclusive because of the dissimilarities across the studies. Besides, humans are exposed to chemical mixtures rather than single compounds, and mixtures can generate numerous effects due to interactions between single compounds (Drakvik et al., 2020). To date, potential chemical mixture effects on gender-specific behavior have not been addressed properly.

Human brain development continues until early adulthood, and studies beyond the prenatal stage have aimed to shed light on the neurobiological underpinnings of gender dysphoria. Neuroimaging studies have identified structural and functional brain differences in connection with gender diversity, and different hypotheses have been proposed to explain how these differences might play a role in gender dysphoria (Gavazzi et al., 2022; Hoekzema et al., 2015; Manzouri & Savic, 2019; Smith et al., 2015; Xerxa et al., 2023).

Psychosocial influences

Hypotheses regarding the involvement of socialization in gender development originated from early work by Mischel (1966) and were further developed through Bussey and Bandura's (1999) application of social cognitive theory to gender development. Succinctly, this framework emphasizes that gendered behaviors develop by observing others and being shaped by feedback received from the external environment. However, TGD children have been shown to prefer toys, clothes, and peers, and to express gendered behaviors in line with their experienced gender (Fast & Olson, 2018; Gülgöz et al., 2019; Olson et al., 2015), despite the evidence showing that parents and peers tend to be more supportive of behaviors aligning with the gender assigned at birth (Lamb et al., 1980; Langlois et al., 1980). Therefore, while acknowledging the impact of social milieu on gender, the socialization perspective alone falls short of explaining gender development among TGD children and youth (deMayo et al., 2022).

Self-cognition in relation to gender, gender identity, and gendered behaviors has also been suggested to play a part in gender identity development (Martin et al., 2002). This perspective underscores an individual's own role in shaping their understanding of gender and associated behaviors, as well as how internal motivations drive gendered expressions (Martin et al., 2002). The emphasis on self-perception of gender, rather than gender solely being influenced by external factors, has been claimed to better explain diverse gender identity development despite external discouragement, as mentioned above (deMayo et al., 2022).

Pathways of gender identity formation

All underlying mechanisms aside, researchers have also studied the trajectories of gender identity formation among TGD individuals. Earlier works most often employed stage models to describe various life stages TGD people experience during their gender identity formation. Devor's (2004) framework which outlines 14 stages until fully embracing diverse gender identity, has been one of the most frequently cited, along with other models (Bockting & Coleman 2007; Lev, 2004).

These stage models have been criticized for various reasons, including putting too much emphasis on medical transition, viewing gender diversity through a binary lens, and suggesting linear and sequential gender identity formation (Kuper et al., 2018; Speechley et al; 2024). In contrast, gender identity formation has lately been conceptualized as a non-linear, flexible, and personalized process by other researchers (Ashley, 2019; Kuper et al., 2018; Pullen Sansfaçon et al., 2020; Riggs, 2019; Speechley et al., 2024). Specifically, Speechley and colleagues (2024) have proposed a diverse gender identity framework by analyzing existing literature. This framework comprises the following themes: a sense of difference, exposure and realization of fit, active gender negotiation, and commitment to gender diverse identity. This model covers both binary and non-binary identities and eliminates medical transition as a fundamental step in gender identity formation.

In addition to the aforementioned models, the role of master narratives, the narratives shared by many within a culture, has also been examined in gender identity formation (Bradford & Syed, 2019; McLean et al., 2017). Bradford and Syed (2019) have shown how transnormative narratives, the narratives that form and demarcate transgender identifications and experiences across different settings, as defined by Johnson (2016), play a significant role in transgender identity formation.

The link between gender nonconformity and gender dysphoria

The relationship between gender nonconformity, particularly during childhood, and gender dysphoria has been subjected to research regarding the developmental perspectives on gender dysphoria. Diagnostic criteria for gender dysphoria and gender incongruence in childhood indicate that gender nonconformity can be one of the manifestations of gender dysphoria or gender incongruence (APA, 2013; WHO, 2019). Indeed, previous studies reported higher gender nonconformity among TGD people in comparison to cisgender people (Koehler et al., 2017; Roshan et al., 2019; Sumia et al., 2017). On the other hand, gender nonconformity has also been shown to be linked to diverse sexual orientations (Li et al., 2017; Marino et al., 2023; Steensma et al., 2013a), but being gender nonconforming does not necessarily indicate sexual or gender variance.

Based on the findings of four studies (Drummond et al., 2008; Steensma et al., 2011; Steensma et al., 2013b; Wallien & Cohen-Kettenis, 2008), it has been suggested that the majority of children referred for gender dysphoria do not continue to experience it later in life, and those who do not experience gender dysphoria have often been associated with diverse sexual orientations. However, concerns regarding the methodology and conceptualization of gender dysphoria in these studies have been raised (Newhook et al., 2018). The intricate relationship between gender nonconformity, diverse sexual orientations, and gender dysphoria is still insufficiently studied.

Mental health

The mental health status of TGD individuals has been one of the most studied areas in the medical literature on gender diversity. TGD people constitute a heterogeneous group, and their mental well-being can vary greatly.

Associations with psychological difficulties

In general, TGD people have been shown to be at a higher risk of psychological difficulties and mental health issues compared to the general population (Reisner et al., 2016). Higher prevalences of depression, anxiety, substance use, self-harm behavior, and suicidality have been found among TGD individuals in comparison to the general population (Bouman et al., 2017; Connolly & Gilchrist, 2020; Davey et al., 2016; Erlangsen et al., 2023; Klinger et al., 2024; Nyquist et al., 2025; Witcomb et al., 2018). Furthermore, TGD people have been reported to experience more traumatic events than cisgender people (Biedermann et al., 2021; Ramos & Marr, 2023; Wirtz et al., 2020). Regardless of being TGD, expressing gender nonconformity alone has also been linked to a heightened risk of mental health problems (Lowry et al., 2018; Narita et al., 2024; Xu et al., 2024).

Although the misalignment between one's gender identity and sex assigned at birth can precipitate distress, being TGD does not per se cause mental health issues. Decreased life quality and mental well-being among TGD people have been linked to discrimination, stigmatization, exclusionary behavior, and traumatic experiences (Başar et al., 2016; Bränström & Pachankis, 2021; Pellicane & Ciesla, 2022). The interconnection between stigma-related stress and adverse mental health consequences among TGD individuals is conventionally explained in terms of the minority stress model (Meyer, 2003). Concisely, having a minority identity may be accompanied by external prejudice-related experiences such as discrimination and violence, as well as internal factors, including an expectation of rejection, concealment, and internalized phobia, all of which can contribute to adverse mental health outcomes (Meyer, 2003).

The model was not designed specifically for TGD people but it has widely been utilized when studying TGD identities (Mezza et al., 2024).

Co-occurrence with autism

Autism spectrum disorder (ASD) is a neurodevelopmental condition which is characterized by deficits in social communication and social interaction, and restricted, repetitive patterns of behavior, interests, or activities (APA, 2013). The prevalence of ASD is estimated at around 1% worldwide, and its underlying causes are considered multifactorial (Lord et al., 2020; Zeidan et al., 2022). There is currently no consensus regarding the terms used to describe autism; research has shown that people may prefer either person-first language (e.g., person with autism) or identity-first language (e.g., autistic person) (Bury et al., 2023; Taboas et al., 2023). In light of the variety in language preferences, I use both person-first and identity-first languages in this thesis.

The high co-occurrence of ASD and TGD identities has been studied extensively over the past 15 years. Increased rates of ASD and autistic traits among TGD people in comparison to the general population or cisgender control groups have been demonstrated in various studies (Becerra-Culqui et al., 2018; David et al., 2025; de Vries et al., 2010; Huisman et al., 2024; Kung, 2020; Mahfouda et al., 2019; Skagerberg et al., 2015; van der Miesen et al., 2018a; Warrier et al., 2020). Associations between gender nonconformity and ASD or related traits have also been documented (Schiltz et al., 2023; Shumer et al., 2015). According to one meta-analysis, the prevalence of ASD among individuals with gender dysphoria and gender incongruence is estimated at 11% (Kallitsounaki & Williams, 2023). Similarly, an increased proportion of gender variance has also been shown among people with ASD (Janssen et al., 2016; van der Miesen et al., 2018b; Strang et al., 2014).

Differing from studies reporting a high co-occurrence of ASD and TGD identities and/or gender dysphoria, few studies have investigated how ASD, autistic traits, and gender dysphoria-related characteristics are interconnected. Fischbach et al. (2024) reported no significant differences in experience of binary or non-binary gender identity and gender dysphoria symptoms between transgender youth with and without ASD. Likewise, among clinically referred youth for gender affirming care in Australia, there were no significant differences in gender identity, gender expression, and gender dysphoria between those with clinically significant autistic traits and those without (Tollit et al., 2024). Similar patterns of gender dysphoria and childhood gender-specific behavior among transgender people with and without ASD were also reported (Kallitsounaki & Williams, 2022). Overall, these studies suggest no association of ASD with gender dysphoria symptoms and gender nonconformity among TGD people.

Various potential underlying mechanisms for the co-occurrence of ASD and gender dysphoria, including biological, psychological, and social factors, have been proposed (van der Miesen et al., 2016). Concerning biological factors, a shared genetic predisposition for both conditions, prenatal exposure to androgens, and common predisposing factors such as low birth weight have been mentioned in the literature (Wattel et al., 2024). Moreover, the extreme male brain (EMB) theory has been proposed to explain the relationship between ASD and gender dysphoria. Briefly, the EMB theory suggests that ASD is linked to a pronounced male cognitive profile, characterized by strong systemizing and weak empathizing (Baron-Cohen, 2002). High prenatal testosterone levels may lead to ASD and influence gender identity development (Baron-Cohen, 2009; Jones et al., 2012). The EMB theory remains controversial, as conflicting findings and criticism have been put forward (Kung et al., 2016; Ridley, 2019; Whitehouse et al., 2012). Regarding psychological explanations, cognitive rigidity, impairments in mentalizing capacity, and potential gender-related obsessions among autistic people have been proposed as contributing factors to gender dysphoria (Wattel et al., 2024). Alongside these factors, difficulties in adhering to social norms, social communication deficits among people with ASD, and other social factors have also been suggested as drivers of the link between ASD and gender dysphoria (Wattel et al., 2024). Thus far, the underlying mechanisms remain elusive.

Even though evidence indicating a relationship between ASD and gender dysphoria has accumulated, discrepancies in findings across studies are noteworthy. This is potentially, at least in part, due to the different tools used to measure ASD, gender dysphoria, and related traits across studies. It has been suggested that previous studies might have misclassified psychological symptoms linked to minority stress among TGD people as autistic traits, which could exaggerate the true association between ASD and gender dysphoria (Fortunato et al., 2022; Turban & van Schalkwyk, 2018). Further research using various tools and investigating different dimensions of ASD and gender dysphoria is warranted.

Research aims

This thesis explores gender nonconformity and gender dysphoria from different angles and aims to broaden the understanding of these phenomena within a multidisciplinary framework. It draws on individual studies conducted within or at the intersections of the fields of psychiatry, epidemiology, gender studies, and biology. The specific research aims are:

- To assess the associations between prenatal exposure to phthalates and gender-specific play behavior (**Paper I**)
- To investigate the associations between gender-specific play behavior and behavioral difficulties and autistic traits (**Paper II**)
- To examine the relationship between gender nonconformity, gender dysphoria, and diverse sexual orientations, as well as the link between gender nonconformity and mental health among people with gender dysphoria (**Paper III**)
- To investigate the co-occurrence of ASD and gender dysphoria, as well as the associations between ASD/autistic traits and gender incongruence in individuals with gender dysphoria (**Paper IV**)
- To explore how people experiencing gender dysphoria reflect on their gender identity formation process and the terms they use to articulate their gender identities (**Paper V**)

Methodology

Cohorts

The individual studies included in this thesis employed three data sources which are explained in detail below.

The Swedish Environmental Longitudinal Mother and Child Asthma and Allergy (SELMA) study

The SELMA study is an ongoing population-based pregnancy cohort study that was established in Värmland, Sweden (Bornehag et al., 2012). The overall aim of the study was to investigate prenatal exposure to EDCs in relation to health outcomes, including asthma and allergy, growth and metabolism, sexual development, and neurodevelopment. Between November 2007 and March 2010, 6,658 pregnant women in the median 10th week of their pregnancy, who spoke Swedish and lived in Värmland, were invited to participate during their antenatal care visits. Of these, 2,582 (39%) consented to participate in the study. The follow-up of the children of these pregnant mothers is still ongoing. Data collection has included biological sampling of urine and blood from the pregnant women and children, environmental sampling of indoor dust and air, parent- and self-reported questionnaires, and physical examinations of children during their routine health check-ups. The SELMA study was approved by the Ethical Review Board in Sweden (Dnr: 2007/062, Dnr: 2015/177, and Dnr: 2024-04849-01). More detailed information about the SELMA study can be found elsewhere (Bornehag et al., 2012).

The Swedish Gender Dysphoria Study (SKDS)

The SKDS is an ongoing multicenter cohort study in Sweden. The overarching aim of the study is to investigate mental health and psychosocial outcomes associated with gender dysphoria, the effects of gender-affirming treatments, and the biological correlates of gender dysphoria. Individuals who were in contact with gender-affirming healthcare, aged 15 years or older, and could speak Swedish, were invited to participate in the SKDS. Recruitment of the participants was conducted at gender clinics in Alingsås/Gothenburg, Linköping, Lund/Malmö, Stockholm, Umeå, and Uppsala. Participants at different stages of their clinical evaluation for gender dysphoria were included in the SKDS. Not all participants had a gender dysphoria diagnosis when they joined the study. However, in this thesis and the respective papers, all participants in contact with gender-affirming healthcare are collectively referred to as "participants with gender dysphoria". Each participant with gender dysphoria has been followed up for three years. Data collection has included self-administered surveys and questionnaires on gender dysphoria and mental health outcomes, neurocognitive assessment, and biological sampling of saliva and deciduous teeth. In addition to participants with gender dysphoria, randomly selected age- and county-matched controls were provided by Statistics Sweden, the governmental agency responsible for Sweden's official statistics. The cisgender control group, composed of individuals without a registered gender dysphoria diagnosis, completed only the baseline assessment of the SKDS. Data linkage to the Swedish National Patient Register which contains primary and secondary ICD diagnoses from healthcare visits was possible for the participants who consented. The SKDS was approved by the Ethical Review Board in Sweden (Dnr: 2016/013), and more detailed information can be found elsewhere (Özel et al., 2023).

Interview participants

For the qualitative study that utilized semi-structured interviews, 16 individuals were recruited between August 2023 and March 2024. The inclusion criteria were set as experiencing gender dysphoria and being at least 15 years of age. The medical diagnosis of gender dysphoria was not a specific inclusion criterion. As the focus of the study was a sensitive topic and participants could be hard to access, they were recruited via snowball sampling (Parker et al., 2019). Information about the study was circulated via university email channels, and participants were either recruited from these email channels or through other participants. All participants provided both oral and written consent to participate in the study. I conducted semi-structured one-on-one interviews with the participants in English; 12 of these were in person, while the other four were via Zoom. The interviews lasted between 48 minutes and 90 minutes (64 minutes on average). The interviews followed an interview guide, which focused on specific thematic concepts, including gender(ed) experiences during childhood and adolescence, gender identity, gender expression, gender dysphoria, the connection between mental well-being and gender dysphoria, and thoughts on research related to gender diversity. This qualitative study was approved by the Ethical Review Board in Sweden (Dnr: 2023-01260-01).

Measures and analytical processes

Paper I

The participants included 715 children at 7.5 years of age from the SELMA study with complete data on prenatal exposure, outcome variables, and covariates.

To measure prenatal phthalate exposure, first morning void urine samples were obtained from pregnant women at enrollment in the median 10th week of their pregnancy. The analyzed phthalate molecules were: di-2-ethylhexyl phthalate (DEHP), di-isononyl phthalate (DiNP), mono-butyl phthalate (MBP), mono-benzyl phthalate (MBzP), mono-ethyl phathalate (MEP), mono-carboxy-isononyl phthalate (MCiNP), mono-hydroxy-isodecyl phthalate (MHiDP), and a phthalate substitute metabolite, 2,4-methyl-7-oxyooctyl-oxycarbonyl-cyclohexane carboxylic acid (MOiNCH). DEHP, a parent phthalate compound, consisted of the molar sum of metabolites of mono-2-ethylhexyl phthalate (MEHP), mono-2-ethyl-5-hydroxyhexyl phthalate (MEHHP), mono-2-ethyl-5-oxohexyl phthalate (MEOHP), mono-2-ethyl-5-carboxypentyl phthalate (MECPP), and mono-2-carboxymethylhexyl phthalate (MCMHP). DiNP, another parent phthalate compound, consisted of the molar sum of metabolites of mono-hydroxy-isononyl phthalate (MHiNP), mono-oxo-isononyl phthalate (MOiNP), and mono-carboxy-isooctyl phthalate (MCiOP). In this thesis, I refer to all chemicals we investigated as phthalates to encompass phthalates, their substitutes, and their metabolites.

Gender-specific play behavior was defined as the outcome variable and measured with a slightly modified 23-item version of the PSAI. The PSAI is a validated parent-rated questionnaire focusing on three areas of play behavior, including types of toys, activities, and child characteristics (Golombok & Rust, 1993a). The questionnaire has 24 items; half of all items are considered "feminine" while the other half is considered "masculine". Each item is rated on a 5-point Likert scale from "1 - never" to "5 - very often". Feminine and masculine scores are determined by summing the respective item scores, with higher values reflecting more gender-specific behavior. Besides, a composite score is calculated using a formula established in a prior standardization study (Golombok & Rust, 1993b). Higher composite scores indicate a greater tendency toward masculine play behavior or a reduced inclination toward feminine play behavior. In our study, we evaluated each questionnaire item to assess its ability to distinguish between girls and boys in our study population. The mean scores for the feminine item "avoiding taking risks" and the masculine item "climbing" did not differ significantly between girls and boys. The subscores were calculated using the remaining 21 items, excluding those two.

The associations between prenatal phthalate exposure and play behavior were examined for the single compounds and their mixture. For single compound analyses, associations between log10-transformed concentrations of each phthalate metabolite and gender-specific play behavior were examined using linear regression analysis. To analyze the associations of prenatal exposure to the phthalate mixture with gender-specific play behavior, weighted quantile sum regression (WQS) was used. WQS is a statistical method used to assess the joint effect of multiple environmental exposures on an outcome (Carrico et al., 2015). WQS uses weighted quantiles to capture the combined effects of exposures and creates a single index, which is then used to model the relationship with the outcome. A key advantage of WQS is that it ranks exposures based on their contribution to the cumulative effect on the outcome (i.e., weights). However, a major limitation of this method is the requirement to specify the direction of associations beforehand. Since the direction of the association between the phthalate mixture and play behavior could not be determined from existing literature, we conducted analyses assuming both possible directions. In order to check the stability and generalizability of the results, repeated holdout validation (RHV) was applied. RHV is a model validation technique where the dataset is randomly split into training and testing sets multiple times (Tanner et al., 2019). For each split, the chemical weights are estimated on the training sets and tested on the testing sets. This process results in a distribution of validated results.

All associations between phthalates and gender-specific play behavior were examined using two models. The crude models were adjusted solely for creatinine concentration, while the adjusted models were also adjusted for child age, maternal age at birth, maternal education level, and parental attitudes toward play behavior. All analyses were stratified by sex.

Paper II

The participants included 718 children, aged 7.5 years, from the SELMA study who had complete data on the variables used in the analyses.

Gender-specific play behavior was used as the independent variable and measured in a similar manner as in **Paper I**, using the PSAI. Behavioral difficulties and autistic traits were defined as the dependent variables and measured with the Strengths and Difficulties Questionnaire (SDQ) and the first version of the Social Responsiveness Scale (SRS), respectively. The SDQ is a validated parent-rated measure consisting of 25 items that assess emotional symptoms, conduct problems, hyperactivity and inattention, peer relationship problems, and prosocial behavior (Goodman, 1997). Responses are rated from 0 to 2; the total score is derived by adding up the four difficulty subscores, ranging from 0 and 40. Higher SDQ score totals indicate greater behavioral difficulties, except for the prosocial behavior subscore. In our study, a cut-off score at the 90th percentile or higher was applied to assess clinically relevant behavioral difficulties. The SRS is also a validated parent-rated scale that includes 65 items on social awareness, social cognition, social communication, social motivation, and restrictive interests and repetitive behaviors (Constantino et al., 2003). Responses are rated from 0 to 3; the total score is calculated by adding up all items, with a range from 0 to 195. Higher scores indicate more autistic traits. Sex-standardized T-scores were used in our study, with cut-offs applied to identify clinically relevant autistic traits.

The associations between PSAI scores and SDQ and SRS scores were first examined using linear regression analysis. Using the cut-off scores for SDQ and SRS, the associations between gender-specific play behavior and clinically relevant behavioral difficulties and autistic traits were analyzed with logistic regression analysis. Similar to **Paper I**, two different models were conducted for all analyses: crude models without covariates and adjusted models controlling for child age, maternal age at birth, maternal education level, and parental attitudes toward play behavior. All analyses were stratified by sex.

Paper III

The participants included 330 individuals with gender dysphoria (mean age of 27 years) and 301 individuals without gender dysphoria (mean age of 28 years) from the SKDS, all of whom had complete information on gender nonconformity.

Childhood gender nonconformity was treated as the independent variable and assessed with the Recalled Childhood Gender Identity/Gender Role Questionnaire (RCGI). The RCGI is a validated self-report questionnaire with 23 items on gender conforming and nonconforming expressions and relations with parents (Zucker et al., 2006). Each item is measured on a five-point Likert scale. In our study, only 18 items which were related to the gender identity and behavior factor of the questionnaire were used. We applied reverse coding to each item to facilitate interpretation, ensuring that higher reversed RCGI scores reflect greater gender nonconformity in **Paper III**. Additionally, some RCGI items included a “non-applicable” response option; these responses were omitted when calculating the average total RCGI score.

In the first set of analyses, gender dysphoria and diverse sexual orientations were defined as the outcome variables. Cisgender participants who did not report being primarily or exclusively attracted to the opposite gender were classified as cisgender nonheterosexual. In the second set of analyses, conducted only among participants with gender dysphoria, gender dysphoria- and mental health-related parameters—including age at onset for gender incongruence, severity of gender dysphoria, experiences of discrimination, traumatic experiences, suicidal thoughts and attempts, and self-harm behaviors—were treated as the outcome variables. Information on these variables was collected through self-reported online surveys.

Regarding statistical analyses, the associations between RCGI scores and gender dysphoria were assessed using logistic regression in the full study sample, with interactions with sex assigned at birth also tested. Subsequently, multinomial logistic regression was conducted to examine associations across three categories: gender dysphoria, diverse sexual orientations, and being cisgender. Next, the relationships between childhood gender nonconformity and gender dysphoria- and mental health-related factors were analyzed using linear and logistic regression. These analyses were performed using three models: one without covariate adjustment, one adjusted for potential confounders (sex assigned at birth, age, and education), and the final one including an interaction term for sex assigned at birth.

Paper IV

This study utilized data from the SKDS. The study sample consisted of 372 individuals with gender dysphoria (mean age of 27 years) and 332 individuals without gender dysphoria (mean age of 28 years), all of whom participated in the baseline evaluation of the SKDS.

Having ASD was defined as having at least one ICD-10 diagnosis of F84.0, F84.1, F84.5, F84.8, or F84.9 in the national patient register between 2010 and 2023. Autistic traits were measured with the Ritvo Autism and Asperger Diagnostic Scale-14 (RAADS-14) and the Autism Spectrum Quotient (AQ). The RAADS-14 is a validated self-report scale consisting of 14 items assessing mentalizing deficits, social anxiety, and sensory reactivity (Eriksson et al., 2013). Responses are rated from 0 to 3, and the total score ranges from 0 to 42. Higher RAADS-14 scores reflect a greater level of autistic traits. The AQ is a validated self-report scale consisting of 50 items assessing social skills, attention switching, attention to detail, communication, and imagination. (Baron-Cohen et al., 2001). Responses are scored as either 0 or 1; the total score ranges from 0 to 50. Higher AQ scores indicate more pronounced autistic traits.

Gender incongruence was used as a self-reported experience and measured with the Transgender Congruence Scale (TCS) (Kozee et al., 2012). The TCS has 12 items and two subscales: appearance congruence and gender identity acceptance. Each item is rated on a scale from 1 to 5, with a total score ranging between 12 and 60. In our study, we reverse-coded each item so that higher scores correspond to more pronounced gender incongruence.

In this study, multiple imputation was employed to handle missing values for the included variables. The prevalence of ASD diagnoses and the mean scores of RAADS-14 and AQ were examined in the gender dysphoria and control groups. Using linear regression analysis, the associations between ASD diagnoses, autistic traits, and gender incongruence among participants with gender dysphoria were investigated. The same three different models as in **Paper III** were used for these analyses.

Paper V

This study included 16 participants aged 15 to 35 who were experiencing gender dysphoria. Ten participants were AFAB, and six were AMAB. Different terms used to articulate gender identities were examined across the interviews.

Reflexive thematic analysis was conducted following Braun and Clarke's (2022) approach. Themes and subthemes related to experiences of exploring gender identity across the lifespan were identified. Specifically, after verbatim transcription of the interviews, the content was read and reviewed multiple times. Initial coding was then performed, followed by discussions and reflections on the codes before finalization. The coding process incorporated both inductive and deductive approaches: it was inductive as codes were derived from the data, and deductive since subject matter knowledge guided the study from conceptualization to theme identification.

Results

Paper I

This study aimed to investigate the associations between prenatal phthalate exposure and gender-specific play behavior. The single compound analyses showed that higher concentrations of DiNP were associated with lower scores of both masculine (β = -1.44; 95% confidence interval [CI] = -2.72, -0.16) and composite (β = -1.43; 95% CI = -2.72, -0.13) play behavior among boys. The same analyses for girls revealed a negative association of MOiNCH concentrations with both feminine (β = -1.59; 95% CI = -2.62, -0.57) and masculine (β= -1.22; 95% CI= -2.14, -0.29) play behavior scores.

In boys, an association was observed between exposure to the mixture of phthalates and lower scores in masculine (β = -0.48; 95% CI = -0.80, -0.16) and composite (β = -0.37; 95% CI = -0.67, -0.08) play behavior. This association was found in models that included the full study sample and assumed negative relationships. No significant association was observed with RHV, even though the association for the masculine score was close to the significance level. In girls, greater prenatal exposure to the phthalate mixture was linked to higher masculine play behavior scores (β = 0.30; 95% CI = 0.01, 0.58) in models that included the full study sample and assumed positive associations. In models incorporating the full study sample and assuming negative associations, greater prenatal exposure to the phthalate mixture was associated with lower feminine play behavior scores among girls (β = -0.33; 95% CI = -0.62, -0.03). Similarly to boys, no significant association was observed using RHV in girls.

Paper II

In this study, we aimed to investigate the associations between gender-specific play behavior and behavioral difficulties as well as autistic traits. The findings on autistic traits indicated that, among girls, higher composite play behavior scores were significantly associated with the SRS total score (β = 0.13; 95% CI = 0.00, 0.26). Significant associations were also shown for the social awareness and social cognition subscales. Additionally, higher feminine play behavior scores were associated with lower odds of exhibiting clinically relevant autistic traits (odds ratio [OR] = 0.90; 95% CI = 0.82, 0.98) when the lower cut-off score was applied. No significant associations with SRS scores were found in boys.

The findings on behavioral difficulties demonstrated that higher composite play behavior scores in girls were associated with increased SDQ total scores ($\beta = 0.11$; 95% CI = 0.04, 0.18). Different significant associations were also observed between gender-specific play behavior scores and conduct problems, hyperactivity and inattention, and prosocial behavior among girls. Moreover, higher feminine play behavior scores (OR = 0.93; 95% CI = 0.87, 0.99) and composite play behavior scores (OR = 1.07; 95% CI = 1.02, 1.13) were linked to the odds of exhibiting clinically relevant behavioral difficulties among girls. Among boys, both masculine ($\beta = 0.10$; 95% CI = 0.02, 0.19) and composite scores ($\beta = 0.10$; 95% CI = 0.01, 0.18) showed positive associations with the total SDQ score. Conduct problems, hyperactivity and inattention, and peer relationship problems were also associated with different play behavior subscores.

Paper III

This study aimed to investigate the links between gender nonconformity, gender dysphoria, and diverse sexual orientations. A significant association was observed between childhood gender nonconformity and gender dysphoria later in life (OR = 14.5; 95% CI = 9.7, 21.5). In the multinomial regression model, using cisgender nonheterosexual individuals as the reference group, greater gender nonconformity was associated with higher odds of gender dysphoria (OR = 6.3; 95% CI = 3.7, 10.5) and lower odds of being cisgender heterosexual (OR = 0.3; 95% CI = 0.2, 0.5).

In individuals with gender dysphoria, the associations between gender nonconformity and mental health were also examined. Among participants with gender dysphoria, our analyses revealed significant associations between gender nonconformity and earlier onset of gender incongruence ($\beta = -4.8$; 95% CI = -5.8, -3.7) as well as greater severity of gender dysphoria ($\beta = 6.8$; 95% CI = 3.2, 10.4). Gender nonconformity was also linked to greater experiences of discrimination ($\beta = -20.0$; 95% CI = 12.4, 27.7), with this association being more pronounced among individuals AMAB than those AFAB. Additionally, positive associations between higher levels of gender nonconformity and exposure to sexual assault (OR = 1.5; 95% CI = 1.0, 2.2) and suicide attempts (OR = 1.6; 95% CI = 1.1, 2.5) were shown.

Paper IV

We aimed to investigate the co-occurrence of gender dysphoria and ASD in this study. Among individuals AFAB, the prevalence of ASD was 24.4% in those with gender dysphoria, compared to 3.6% in those without. For individuals AMAB, 21.5% of those with gender dysphoria had an ASD diagnosis, whereas the prevalence was 2.0% among those without. Overall, the risk of having an ASD diagnosis was 7.8 times higher in individuals with gender dysphoria than in those without (95% CI = 3.9, 15.3).

Participants with gender dysphoria reported significantly higher total scores on the RAADS-14, among both individuals AFAB (mean difference [MD] = 10.1; 95% CI = 8.2, 12.1) and those AMAB (MD = 5.1; 95% CI = 2.8, 7.5). Both participants AFAB and AMAB with gender dysphoria scored higher on all RAADS-14 subscales in comparison to their cisgender counterparts. When comparing individuals with and without gender dysphoria, only participants AFAB—but not participants AMAB—had significantly higher AQ total scores (MD = 7.5; 95% CI = 5.9, 9.2). Findings regarding the AQ subscales were mixed in terms of differences between groups.

Regarding associations with self-reported experiences of gender incongruence, a significant association was observed only for the total AQ score among participants with gender dysphoria ($\beta = 0.09$; 95% CI = 0.00, 0.18).

Paper V

This study aimed to explore the terms used to describe gender identities and experiences throughout the formation of gender identity among people experiencing gender dysphoria. Our results showed that participants often used multiple terms simultaneously for their gender identifications, and these terms evolved throughout their exploration of their gender identity. Their choice of terms was influenced by multiple factors, including for example parental attitudes during childhood and their country of origin. Furthermore, participants used similar terms in different ways, which was particularly evident in the use of the term trans.

The first main theme in the data was identified as “gender awareness during childhood” with two subthemes “early awareness of gender identity” and “lack of gender identity awareness”. The second main theme was “gender discovery during adolescence and early adulthood”, with two subthemes: “initial lack of knowledge about gender diversity” and “internal conflicts about gender identity”. The last theme was “landing on a gender identity”, consisting of “non-binary identification” and “genderless self-perception”. Overall, our participants had varying degrees of awareness of their gender identity during their early years. Some were unfamiliar with the concept of gender diversity, while others struggled with internal conflicts during their adolescent and early adult years. Ultimately, most participants identified as non-binary and/or viewed themselves as genderless rather than adopting binarist terms.

Discussion

The overarching aim of this thesis is to enhance our understanding of gender nonconformity and gender dysphoria with the help of different perspectives. The individual studies included in this thesis examine gender nonconformity and/or gender dysphoria from different viewpoints, depending on their respective disciplines, and employ varied methodological approaches. The findings of each study are discussed in detail in the individual papers. In this section, I aim to provide a broader discussion of the specific aspects of the two phenomena in focus by integrating insights from the different studies.

Endocrine disrupting chemicals in the context of gender nonconformity and gender dysphoria

In **Paper I**, we found negative associations between prenatal DiNP exposure and masculine and composite play behavior scores in boys, and negative associations between prenatal MOiNCH exposure and feminine and masculine play behavior scores in girls. Our analyses regarding the phthalate mixture suggested links between prenatal phthalate exposure and gender-specific play behavior in both girls and boys. Overall, our findings support an association between prenatal phthalate exposure and gender-specific play behavior, consistent with previous studies (Evans et al., 2021; Liu et al., 2023; Percy et al., 2016; Swan et al., 2010). Differences in contributions of phthalate metabolites across studies may be partly explained by variations in phthalate concentrations and methodological differences.

The mechanisms underlying the associations described in **Paper I** remain speculative. However, the observed associations may, in part, be attributed to phthalates' potential to interfere with sex hormone signaling. In particular, phthalates have been shown to act as anti-androgens, i.e. inhibiting various aspects of testosterone action (Czernych et al., 2017; Zhu et al., 2022). Testosterone is thought to induce long-lasting and stable effects on the developing brain, known as organizational effects, while also modulating neural circuits later in life through activating effects (Hines, 2020). Increased prenatal exposure to testosterone has been associated with greater masculinity in girls (Meyer-Bahlburg et al., 2006; Nordenström et al., 2002; Pasterski et al., 2011; Pasterski et al., 2015; Servin et al., 2003), while boys who were not sensitive to androgens tended to display more feminine traits (Hines et al., 2003; Jürgensen et al., 2007). Thus, the well-described anti-androgenic effects of phthalates can potentially explain the associations observed in boys. In girls, however, the associations were more mixed, as exemplified by MOiNCH being associated with both decreased feminine and masculine play behavior. This could be due to phthalates' capacity, in addition to their anti-androgenic properties, to interact with various sex hormone receptors, leading to diverse hormonal disruptions (Aimuzi et al., 2022; Hlisníková et al., 2020). For instance, MOiNCH has been shown to have androgenic, estrogenic, and anti-estrogenic effects (Engel et al., 2018), which might have led to the differing associations with gender-specific play behavior in girls in **Paper I**.

In addition to the role of prenatal androgen exposure in brain development and sexual differentiation, Daphna Joel's (2021) mosaic brain hypothesis also offers an important perspective for interpreting our findings. According to this hypothesis, while sex differences in the brain exist at the group level, there is no such thing as a sex-typical brain at the individual level. This should be considered when interpreting the differing associations in our study, particularly when considered in the context of brain feminization or masculinization.

Our results should not be interpreted through the lens of biological determinism. Although **Paper I** focused on examining potential biological underpinnings of gender nonconforming play, sociocultural aspects of play behavior were acknowledged throughout the research process. Associations of certain toys and play styles with specific genders are shaped within certain sociocultural contexts, vary across cultures, and are likely changing over time. Gender-specific play behavior was not conceptualized as being solely or inherently biological in our study. However, our study, based on a Swedish population from the 2010s, found significant differences in gender-specific play behavior between girls and boys. Thus, we explored one potential contributor to variations in these distinct play behavior patterns.

As outlined in detail in the introduction, a potential relationship between EDCs and gender dysphoria has been proposed in the literature, but concrete evidence is lacking. In **Paper I**, associations between EDC exposure and gender-specific play behavior, not for gender dysphoria, were shown. On the other hand, in **Paper III**, we showed positive associations between childhood gender nonconformity and gender dysphoria. This could imply a link between prenatal EDC exposure and gender dysphoria. However, childhood gender nonconformity was also shown to be linked to diverse sexual orientations, although the associations were more pronounced for gender dysphoria. While

Paper I and **III**, when considered together, may suggest indirect associations with gender dysphoria, the findings of **Paper I** should not be extrapolated in relation to gender identity or gender dysphoria. **Paper I** focused on only one dimension of gender nonconformity—gender-specific play behavior—whereas **Paper III** examined different representations of gender nonconformity using a different scale in a different study population. Without directly addressing EDC exposure and gender dysphoria in the same study, suggesting a link between EDCs and gender dysphoria would be too speculative. Additionally, as we exemplified in **Paper V**, individuals experiencing gender dysphoria have diverse experiences regarding how they perceive their gender identity and express gender nonconformity during childhood. Inferring gender dysphoria solely based on childhood gender nonconformity would oversimplify the range of experiences among TGD individuals and could be misleading.

Studying the associations between prenatal EDC exposure and gender-specific play behavior offers two key benefits. First, it can enhance our understanding of the impact of EDCs on human behavior. More importantly, and in line with the aim of this thesis, it may provide valuable insights into the development of gendered behavior, gender identity, gender nonconformity, and, consequently, gender diversity. Ultimately, this biological evidence could inform practices such as gender-affirming hormone treatments. The results of this thesis provide a scientific foundation for future biomedical research in the field. For instance, studying prenatal androgen exposure could help clarify the neurobiological correlates of gender identity. However, like many other scientific findings, biological research on gender nonconformity and gender dysphoria is open to misinterpretation. Biomedical research in this field should focus on improving people's lives without contributing to further stigmatization (Kennis et al., 2024). To ensure this, engaging with advisory boards consisting of TGD individuals and conducting multidisciplinary research on clinically relevant issues, such as gender-affirming treatments, have been suggested (Kennis et al., 2024).

Implications of gender nonconformity for mental well-being

Studying gender nonconformity is important because it is linked to discrimination and stigmatization which can negatively impact mental well-being (Bränström et al., 2023; Lian et al., 2022; Narita et al., 2024; Oginni et al., 2019; Zhao et al., 2021). **Paper II** scrutinized gender-specific play behavior in children, while **Paper III** examined broader representation of childhood gender nonconformity.

In **Paper II**, increased masculinity in play was associated with behavioral difficulties, particularly hyperactivity and conduct problems, in both girls and boys. The associations observed in girls may be driven by the impact of gender nonconformity on behavioral outcomes, as suggested in previous literature (van Beijsterveldt et al., 2006; van der Miesen et al., 2018c). However, in boys the associations cannot be interpreted from the point of gender nonconformity. It is possible that masculine play behavior was misclassified as behavioral difficulties. Furthermore, increased feminine play behavior in boys was associated with peer relationship problems in **Paper II**. The potential role of prejudice and stigmatization toward boys engaging in more feminine play may have contributed to this relationship. This finding from **Paper II** aligns with some of the results in **Paper III**. Specifically, greater childhood gender nonconformity was associated with increased experiences of discrimination among people with gender dysphoria in **Paper III**, with a stronger association for participants AMAB. These findings from **Paper II** and **III** are consistent with the existing literature suggesting lower tolerance for gender nonconformity in AMAB in comparison to AFAB (Conry-Murray et al., 2015; Spivey et al., 2018; van Beusekom et al., 2020). Taken together, our results may indicate a heightened risk of discrimination, and consequently poorer mental well-being, for gender nonconforming people AMAB compared to AFAB in certain contexts.

Although the connection between gender nonconformity and mental health is well-established in the literature, the impact of gender nonconformity on mental health specifically among individuals with gender dysphoria has been less frequently studied. Previous qualitative studies conducted with TGD individuals have shown how gender(ed) expressions can influence daily life and sometimes lead to distress due to the reactions of others (Connell, 2010; Levitt & Ippolito, 2013). Additionally, a link between gender nonconformity, discrimination, and self-harming behavior among TGD people has been shown (Miller & Grollman, 2015). **Paper III** yielded similar results, suggesting more severe gender dysphoria as well as increased exposure to sexual assault and higher odds of suicide attempts for people with gender dysphoria who exhibited greater gender nonconformity. As Miller and Grollman (2015) discussed, these associations may be interpreted in relation to stigma visibility, where individuals with visible minority status are more prone to stigmatization. Being gender nonconforming might intensify the likelihood of facing discrimination and result in adverse life events for people with gender dysphoria.

Notably, the cross-sectional study design in both **Paper II** and **III** prevents us from drawing causal conclusions. The small effect sizes, particularly in **Paper II**, should also be taken into consideration when interpreting our results.

Links between gender nonconformity, gender dysphoria, and autism

This thesis also explored the widely debated co-occurrence of TGD identities and ASD in two different contexts. First, **Paper II** examined gender-nonconforming play behavior and autistic traits in children from a non-clinical population. Second, **Paper IV** focused on ASD diagnoses and autistic traits in individuals seeking gender-affirming healthcare. Utilizing both clinical and non-clinical study populations offers distinct advantages, and focusing on different age groups with various scales may contribute to a better understanding of this co-occurrence.

Paper II suggested increased autistic traits in girls who exhibited more gender nonconforming play, as indicated by higher composite PSAI scores. **Paper IV** confirmed the high co-occurrence of gender dysphoria and ASD by using different autism measures and clearly demonstrated increased prevalence of ASD diagnoses and higher autistic trait scores among people with gender dysphoria compared to their cisgender counterparts. Moreover, participants AFAB with gender dysphoria exhibited more pronounced autistic traits than AMAB. Taken together, **Paper II** and **IV** suggest stronger associations of gender nonconformity and gender dysphoria with autism in individuals AFAB. The lack of significant results in **Paper II** compared to **Paper IV** was expected, as **Paper II** relied on proxy variables, such as play behavior and SRS scores in a population-based sample, whereas **Paper IV** used specific diagnoses of gender dysphoria and ASD. Additionally, despite the associations observed in **Paper III**, using gender-specific play behavior to infer conclusions about gender dysphoria would be inappropriate, as previously discussed. Nonetheless, our results collectively indicate more pronounced associations with autism for individuals AFAB.

Several hypotheses have been proposed to explain the co-occurrence of gender dysphoria and autism (van der Miesen et al., 2016). As outlined in the introduction, the EMB theory links ASD to a male cognitive profile with strong systemizing and weak empathizing and suggests that elevated prenatal testosterone may contribute to ASD and influence gender identity (Baron-Cohen, 2002; Jones et al., 2012). The EMB theory has frequently been cited in the literature, and our results from **Paper II** and **IV**, suggesting stronger associations for individuals AFAB, align with the EMB theory. Yet, our findings do not provide direct support for the EMB theory, as **Paper II** and **IV** lacked biological measures of prenatal testosterone exposure. Moreover, we also found significant associations for individuals AMAB in **Paper IV**, which cannot be explained in the EMB theory framework. The reasons behind this high co-occurrence and stronger associations reported for individuals

AFAB in some studies (Cooper et al., 2018; David et al., 2025; Hendriks et al., 2022; Kung, 2020; Nobili et al., 2018), including our own, remain speculative.

Gender dysphoria has also been suggested to be a result of having ASD based on various hypotheses (Wattel et al., 2024); however, robust evidence supporting this claim is lacking. In **Paper IV**, we did not find any significant association between having ASD and experience of gender incongruence among people with gender dysphoria. In this regard, our results do not support the notion that gender dysphoria is a manifestation of ASD, though further investigations are warranted on this issue.

Examining the link between gender dysphoria and ASD from different perspectives is critical, as people with both conditions might face greater psychological difficulties and barriers in accessing healthcare in comparison to those with only one of these conditions (Strang et al., 2023; Strauss et al., 2021). Clinical recommendations for assessing the intersection of gender dysphoria and autism have been proposed (Dewinter et al., 2024; Strang et al., 2018). Further studies aimed at improving healthcare for these groups are required.

Varieties of gender identity formations

The first four studies included in this thesis investigated gender nonconformity and/or gender dysphoria using quantitative methods in two cohort studies. On the other hand, **Paper V** aimed to gain in-depth knowledge about the experiences of people with gender dysphoria during their gender identity formation processes. This provided enriched information at an individual level without having the aim of generalizing.

The interviews I conducted provided insights into the trajectories of gender identity formation among people with gender dysphoria. Using a qualitative approach offered different advantages compared to quantitative analyses, complementing the overall findings on gender nonconformity and gender dysphoria. For instance, **Paper III** revealed that different gender nonconforming behavior and expressions during childhood were associated with gender dysphoria to varying degrees. In addition to this, **Paper V** highlighted the variety of experiences in relation to gender identity during childhood, emphasizing the individuality of experiences and the heterogeneous nature of TGD individuals as a group. Therefore, generalizing normative definitions should be avoided when describing gender identifications of people with gender dysphoria.

The majority of the participants identified as non-binary and/or genderless in **Paper V**. Even though this finding cannot be generalized to everyone with gender dysphoria, it seems to be in line with current discourses on gender diversity. These terms, increasingly used over the past two decades to define gender identities, should be taken into account in studies seeking to capture information on gender identity. **Paper V** also demonstrated that people experiencing gender dysphoria used multiple terms to describe their gender identities, with the same term sometimes carrying different meanings. This again underscores the individualistic nature of people's experiences. Furthermore, it calls researchers' attention to the need to be explicit about the meaning of the terminology they use, with specific definitions that prevent arbitrary generalizations.

Overall, **Paper V** provides valuable individual-level information to better understand the two phenomena, gender nonconformity and gender dysphoria, in addition to other studies that address these issues at the group level.

Conclusion

This thesis adopted a multidisciplinary approach to explore gender nonconformity and gender dysphoria from various perspectives. Given that gender identity development and emergence of gender dysphoria are multifactorial processes influenced by biological, psychological, and sociocultural factors, this multidisciplinary design was essential and valuable for this research. In summary, this thesis contributes several scientific findings: biologically, prenatal phthalate exposure is linked to gender nonconforming play. Childhood gender nonconformity is associated with gender dysphoria, while gender identity formation is a dynamic and individual process. Gender nonconformity may also increase the risk of decreased mental well-being in individuals with gender dysphoria. Finally, suggestive associations between gender nonconforming play and autistic traits are observed during childhood, particularly for girls, with a notable co-occurrence of gender dysphoria and ASD later in life, which is also more pronounced for individuals AFAB. Overall, this thesis provides valuable insights into different dimensions of gender nonconformity and gender dysphoria.

Svensk Sammanfattning

Under de senaste åren har frågor om könsidentitet och könsmångfald blivit alltmer omdiskuterade, både inom forskningen och i samhällsdebatten. Detta ökade intresse har även riktats mot fenomen som könsöverskridande uttryck och könsdysfori, vilka utgör centrala teman i denna avhandling. Könsöverskridande uttryck innebär att en persons sätt att uttrycka sitt kön (exempelvis genom klädstil, beteende eller kroppsspråk) avviker från de normer och förväntningar som traditionellt kopplas till det kön som tilldelades vid födseln. Könsdysfori handlar däremot om det psykiska lidande som kan uppstå vid en betydande diskrepans mellan en individs könsidentitet och det tilldelade könet. I Sverige, liksom i flera andra länder, har antalet könsdysforidiagnoser ökat under de senaste åren, särskilt bland personer som tilldelades kvinnligt kön vid födseln. Oavsett orsakerna bakom denna utveckling är det tydligt att en fördjupad förståelse för könsidentitet och könsdysfori är av stor betydelse.

Könsidentitetsutveckling är en komplex process som påverkas av biologiska, psykologiska och sociala faktorer. Ur ett biologiskt perspektiv har forskning visat att könshormoner som påverkar hjärnans utveckling under fosterlivet spelar en avgörande roll. Studier har visat att högre nivåer av testosteron under fosterstadiet är kopplade till mer maskulinitet, medan minskad känslighet för testosteron kan förknippas med mer femininitet. Dessa rön har lett till teorier om att hormonstörande ämnen – substanser som kan påverka kroppens hormonella system – möjligen kan ha en inverkan på könsidentitetsutvecklingen. Dock saknas ännu konkreta belägg för ett sådant samband. Psykologiska och sociala faktorer, såsom socialisering och individens kognitiva egenskaper, har också betydelse för hur könsidentiteten utvecklas. Forskning har dessutom visat att könsidentitetsutvecklingen hos transpersoner är en mycket varierande och individuell process. Vidare har studier pekat på samband mellan könsöverskridande uttryck under barndomen och senare könsdysfori, men även på kopplingar mellan sådana uttryck och olika sexuella läggningar. Den invecklade relationen mellan könsöverskridande uttryck, könsdysfori och olika sexuella läggningar är dock fortfarande inte klarlagt.

Den psykiska hälsan hos transpersoner har undersökts ingående i forskningen, och resultaten visar att denna grupp löper en ökad risk för psykisk ohälsa jämfört med övriga befolkningen. Flera studier rapporterar en högre förekomst av depression, ångest, självskadebeteende och suicid bland transpersoner. Även könsöverskridande uttryck i sig har kopplats till en ökad risk för psykiska besvär, men sambandet mellan könsöverskridande uttryck och psykisk hälsa bland transpersoner är fortfarande otillräckligt utforskat. Ett annat forskningsområde som har fått ökad uppmärksamhet är sambandet mellan könsdysfori och autism. Under de senaste 15 åren har flera studier rapporterat en högre förekomst av autism och autistiska drag bland transpersoner. Resultaten är dock blandade, och metodologiska begränsningar har lyfts fram i forskningslitteraturen. De bakomliggande orsakerna till sambandet mellan autism och könsdysfori är ännu oklara, vilket kräver vidare forskning för att bättre förstå detta komplexa samband.

Målet med denna avhandling var att undersöka könsöverskridande uttryck och könsdysfori ur olika perspektiv och att bredda förståelsen av dessa fenomen inom en tvärvetenskaplig ram. Denna avhandling bygger på data från två större studier: SELMA-studien, som undersöker hur miljöfaktorer påverkar barns och mödrars hälsa, samt Svenska Könsdysforistudien (SKDS), som följer personer med könsdysfori i Sverige. Utöver dessa datakällor genomfördes även intervjuer med personer som upplever könsdysfori. Resultaten visar flera centrala samband som bidrar till en fördjupad förståelse av könsöverskridande uttryck och könsdysfori. Prenatal exponering för ftalater – en grupp kemikalier med potentiellt hormonstörande effekter – visade sig vara kopplad till könsöverskridande lekbeteende i barndomen. Dessutom identifierades samband mellan könsöverskridande lekbeteende och beteendesvårigheter samt autistiska drag, särskilt hos flickor.

Vidare framkom att könsöverskridande uttryck i barndomen var associerade med både könsdysfori och olika sexuella läggningar, där sambandet var särskilt starkt för könsdysfori. Högre grad av könsöverskridande uttryck var även kopplat till negativa psykiska hälsoeffekter bland personer med könsdysfori. Resultaten visade dessutom att personer med könsdysfori oftare hade autism eller mer uttalade autistiska drag jämfört med cispersoner (personer som identifierar sig med det kön de tilldelades vid födseln), särskilt bland dem som tilldelades kvinnligt kön vid födseln. Däremot fanns inget tydligt samband mellan att ha en autismdiagnos och att själv uppleva könsinkongruens. Slutligen visade studierna att könsidentitetsutvecklingen hos personer med könsdysfori är en dynamisk och icke-linjär process, med stor variation i hur individer beskriver och uppfattar sin könsidentitet.

Genom att kombinera biologiska, psykologiska och sociala perspektiv belyser denna avhandling flera dimensioner av könsöverskridande uttryck och könsdysfori och bidrar därmed till en bredare och djupare förståelse för dessa komplexa och mångfacetterade fenomen.

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References

Åhs, J. W., Dhejne, C., Magnusson, C., Dal, H., Lundin, A., Arver, S., Dalman, C., & Kosidou, K. (2018). Proportion of adults in the general population of Stockholm County who want gender-affirming medical treatment. *PLOS One*, *13*(10), e0204606.

Aimuzi, R., Wang, Y., Luo, K., & Jiang, Y. (2022). Exposure to phthalates, phenols, and parabens mixture and alterations in sex steroid hormones among adolescents. *Chemosphere*, *302*, 134834.

Aitken, M., Steensma, T. D., Blanchard, R., VanderLaan, D. P., Wood, H., Fuentes, A., Spegg, C., Wasserman, L., Ames, M., Fitzsimmons, C. L., Leef, J. H., Lishak, V., Reim, E., Takagi, A., Vinik, J., Wreford, J., Cohen-Kettenis, P. T., de Vries, A. L., Kreukels, B. P., & Zucker, K. J. (2015). Evidence for an altered sex ratio in clinic-referred adolescents with gender dysphoria. *The Journal of Sexual Medicine*, *12*(3), 756–763.

American Psychiatric Association. (2000). *Diagnostic and statistical manual of mental disorders* (4th ed., text rev.). American Psychiatric Association.

American Psychiatric Association. (2013). *Diagnostic and statistical manual of mental disorders* (5th ed.). American Psychiatric Association.

American Psychological Association. (2015). Guidelines for psychological practice with transgender and gender nonconforming people. *American Psychologist, 70*(9), 832–864.

Arnold A. P. (2009). The organizational-activational hypothesis as the foundation for a unified theory of sexual differentiation of all mammalian tissues. *Hormones and Behavior*, *55*(5), 570–578.

Ashley, F. (2019). Thinking an ethics of gender exploration: against delaying transition for transgender and gender creative youth. *Clinical Child Psychology and Psychiatry*, *24*(2), 223–236.

Auyeung, B., Baron-Cohen, S., Ashwin, E., Knickmeyer, R., Taylor, K., Hackett, G., & Hines, M. (2009). Fetal testosterone predicts sexually differentiated childhood behavior in girls and in boys. *Psychological Science*, *20*(2), 144–148.

Baron-Cohen S. (2002). The extreme male brain theory of autism. *Trends in Cognitive Sciences*, *6*(6), 248–254.

Baron-Cohen S. (2009). Autism: the empathizing-systemizing (E-S) theory. *Annals of the New York Academy of Sciences*, *1156*, 68–80.

Baron-Cohen, S., Wheelwright, S., Skinner, R., Martin, J., & Clubley, E. (2001). The Autism-Spectrum Quotient (AQ): evidence from Asperger syndrome/high-functioning autism, males and females, scientists and mathematicians. *Journal of Autism and Developmental Disorders*, *31*(1), 5–17.

Başar, K., Öz, G., & Karakaya, J. (2016). Perceived discrimination, social support, and quality of life in gender dysphoria. *The Journal of Sexual Medicine*, *13*(7), 1133–1141.

Becerra-Culqui, T. A., Liu, Y., Nash, R., Cromwell, L., Flanders, W. D., Getahun, D., Giammattei, S. V., Hunkeler, E. M., Lash, T. L., Millman, A., Quinn, V. P., Robinson, B., Roblin, D., Sandberg, D. E., Silverberg, M. J., Tangpricha, V., & Goodman, M. (2018). Mental health of transgender and gender nonconforming youth compared with their peers. *Pediatrics*, *141*(5), e20173845.

Bejerot, S., Humble, M. B., & Gardner, A. (2011). Endocrine disruptors, the increase of autism spectrum disorder and its comorbidity with gender identity disorder - a hypothetical association. *International Journal of Andrology*, *34*(5 Pt 2), e350.

Biedermann, S. V., Asmuth, J., Schröder, J., Briken, P., Auer, M. K., & Fuss, J. (2021). Childhood adversities are common among trans people and associated with adult depression and suicidality. *Journal of Psychiatric Research*, *141*, 318–324.

Bockting, W. O., & Coleman, E. (2007). Developmental stages of the transgender coming out process: toward an integrated identity. In R. Ettner, S. Monstrey, & A. E. Eyler (Eds.), *Principles of transgender medicine and surgery* (pp. 185–208). Routledge.

Bornehag, C. G., Moniruzzaman, S., Larsson, M., Lindström, C. B., Hasselgren, M., Bodin, A., von Kobyletzkic, L. B., Carlstedt, F., Lundin, F., Nånberg, E., Jönsson, B. A., Sigsgaard, T., & Janson, S. (2012). The SELMA study: a birth cohort study in Sweden following more than 2000 mother-child pairs. *Paediatric and Perinatal Epidemiology*, *26*(5), 456–467.

Bouman, W. P., Claes, L., Brewin, N., Crawford, J. R., Millet, N., Fernandez-Aranda, F., & Arcelus, J. (2017). Transgender and anxiety: a comparative study between transgender people and the general population. *International Journal of Transgenderism*, *18*(1), 16–26.

Bradford, N. J., & Syed, M. (2019). Transnormativity and transgender identity development: a master narrative approach. *Sex Roles*, *81*(5), 306–325.

Braun, V., & Clarke, V. (2022). Conceptual and design thinking for thematic analysis. *Qualitative Psychology, 9*(1), 3–26.

Bränström, R., Fellman, D., & Pachankis, J. (2023). Structural stigma and sexual minority victimization across 28 countries: the moderating role of gender, gender nonconformity, and socioeconomic status. *Journal of Interpersonal Violence*, *38*(3-4), 3563–3585.

Bränström, R., & Pachankis, J. E. (2021). Country-level structural stigma, identity concealment, and day-to-day discrimination as determinants of transgender people's life satisfaction. *Social Psychiatry and Psychiatric Epidemiology*, *56*(9), 1537–1545.

Bury, S. M., Jellett, R., Spoor, J. R., & Hedley, D. (2023). "It defines who I am" or "It's something I have": What language do [autistic] Australian adults [on the autism spectrum] prefer?. *Journal of Autism and Developmental Disorders*, *53*(2), 677–687.

Bussey, K., & Bandura, A. (1999). Social cognitive theory of gender development and differentiation. *Psychological Review*, *106*(4), 676–713.

Byne, W., Karasic, D. H., Coleman, E., Eyler, A. E., Kidd, J. D., Meyer-Bahlburg, H. F. L., Pleak, R. R., & Pula, J. (2018). Gender dysphoria in adults: an overview and primer for psychiatrists. *Transgender Health*, *3*(1), 57–70.

Carrico, C., Gennings, C., Wheeler, D. C., & Factor-Litvak, P. (2015). Characterization of weighted quantile sum regression for highly correlated data in a risk analysis setting. *Journal of Agricultural, Biological, and Environmental Statistics*, *20*(1), 100–120.

Claesdotter-Knutsson, E., Andersson, M. J., Cervin, M., Delfin, C., Håkansson, A., & Broman, N. (2024). Rise in gender dysphoria diagnoses and legal gender changes in Sweden: 2005-2017. *Archives of Sexual Behavior*, *53*(10), 3731–3738.

Coleman, E., Radix, A. E., Bouman, W. P., Brown, G. R., de Vries, A. L. C., Deutsch, M. B., Ettner, R., Fraser, L., Goodman, M., Green, J., Hancock, A. B., Johnson, T. W., Karasic, D. H., Knudson, G. A., Leibowitz, S. F., Meyer-Bahlburg, H. F. L., Monstrey, S. J., Motmans, J., Nahata, L., … Arcelus, J. (2022). Standards of care for the health of transgender and gender diverse people, version 8. *International Journal of Transgender Health*, *23*(Suppl 1), S1–S259.

Connell, C. (2010). Doing, undoing, or redoing gender?: learning from the workplace experiences of transpeople. *Gender & Society, 24*(1), 31–55.

Connolly, D., & Gilchrist, G. (2020). Prevalence and correlates of substance use among transgender adults: a systematic review. *Addictive Behaviors*, *111*, 106544.

Conry-Murray, C., Kim, J. M., & Turiel, E. (2015). Judgments of gender norm violations in children from the United States and Korea. *Cognitive Development, 35,* 122–136.

Constantino, J. N., Davis, S. A., Todd, R. D., Schindler, M. K., Gross, M. M., Brophy, S. L., Metzger, L. M., Shoushtari, C. S., Splinter, R., & Reich, W. (2003). Validation of a brief quantitative measure of autistic traits: comparison of the social responsiveness scale with the autism diagnostic interview-revised. *Journal of Autism and Developmental Disorders*, *33*(4), 427–433.

Cooper, K., Smith, L. G. E., & Russell, A. J. (2018). Gender identity in autism: sex differences in social affiliation with gender groups. *Journal of Autism and Developmental Disorders*, *48*(12), 3995–4006.

Czernych, R., Chraniuk, M., Zagożdżon, P., & Wolska, L. (2017). Characterization of estrogenic and androgenic activity of phthalates by the XenoScreen YES/YAS in vitro assay. *Environmental Toxicology and Pharmacology*, *53*, 95–104.

Davey, A., Arcelus, J., Meyer, C., & Bouman, W. P. (2016). Self-injury among trans individuals and matched controls: prevalence and associated factors. *Health & Social Care in the Community*, *24*(4), 485–494.

David, L. W., Stenberg, N., Diseth, T. H., Helverschou, S. B., Nyquist, C. B., Øien, R. A., & Waehre, A. (2025). Autistic characteristics in a nationally representative clinical sample of adolescents seeking medical gender-affirming treatment in Norway. *Journal of Autism and Developmental Disorders*, *55*(1), 147–157.

de Vries, A. L., Kreukels, B. P., Steensma, T. D., & McGuire, J. K. (2014). Gender identity development: a biopsychosocial perspective. In B. P. Kreukels, T. D. Steensma, & A. L. de Vries (Eds.), *Gender dysphoria and disorders of sex development: progress in care and knowledge* (pp. 53–80). Springer.

de Vries, A. L., Noens, I. L., Cohen-Kettenis, P. T., van Berckelaer-Onnes, I. A., & Doreleijers, T. A. (2010). Autism spectrum disorders in gender dysphoric children and adolescents. *Journal of Autism and Developmental Disorders*, *40*(8), 930–936.

Delli Paoli, E., Di Chiano, S., Paoli, D., Lenzi, A., Lombardo, F., & Pallotti, F. (2023). Androgen insensitivity syndrome: a review. *Journal of Endocrinological Investigation*, *46*(11), 2237–2245.

deMayo, B. E., Jordan, A. E., & Olson, K. R. (2022). Gender development in gender diverse children. *Annual Review of Developmental Psychology*, *4*, 207–229.

Dessens, A. B., Cohen-Kettenis, P. T., Mellenbergh, G. J., vd Poll, N., Koppe, J. G., & Boer, K. (1999). Prenatal exposure to anticonvulsants and psychosexual development. *Archives of Sexual Behavior*, *28*(1), 31–44.

Devor, A. H. (2004). Witnessing and mirroring: a fourteen stage model of transsexual identity formation. *Journal of Gay & Lesbian Psychotherapy*, *8*(1-2), 41–67.

Dewinter, J., Onaiwu, M. G., Massolo, M. L., Caplan, R., van Beneden, E., Brörmann, N., Crehan, E. T., Croen, L. A., Faja, S., Gassner, D. L., Graham Holmes, L., Hughes, C., Hunter, M., Huysamen, M., Jelonche, P., Lai, M. C., Noens, I., Pukki, H., Stokes, M. A., … van der Miesen, A. I. (2024). Short report: recommendations for education, clinical practice, research, and policy on promoting well-being in autistic youth and adults through a positive focus on sexuality and gender diversity. *Autism: The International Journal of Research and Practice*, *28*(3), 770–779.

Diamond, M. (2013). Transsexuality among twins: identity concordance, transition, rearing, and orientation. *International Journal of Transgenderism, 14*(1), 24–38.

Drakvik, E., Altenburger, R., Aoki, Y., Backhaus, T., Bahadori, T., Barouki, R., Brack, W., Cronin, M. T. D., Demeneix, B., Hougaard Bennekou, S., van Klaveren, J., Kneuer, C., Kolossa-Gehring, M., Lebret, E., Posthuma, L., Reiber, L., Rider, C., Rüegg, J., Testa, G., … Bergman, Å. (2020). Statement on advancing the assessment of chemical mixtures and their risks for human health and the environment. *Environment International*, *134*, 105267.

Drummond, K. D., Bradley, S. J., Peterson-Badali, M., & Zucker, K. J. (2008). A follow-up study of girls with gender identity disorder. *Developmental Psychology*, *44*(1), 34–45.

Engberg, H., Nordenström, A., & Hirschberg, A. L. (2025). Clinical manifestations and challenges in adolescent and adult females with classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *The Journal of Clinical Endocrinology and Metabolism*, *110*(Supplement_1), S37–S45.

Engel, A., Buhrke, T., Kasper, S., Behr, A. C., Braeuning, A., Jessel, S., Seidel, A., Völkel, W., & Lampen, A. (2018). The urinary metabolites of DINCH® have an impact on the activities of the human nuclear receptors ERα, ERβ, AR, PPARα and PPARγ. *Toxicology Letters*, *287*, 83–91.

Eriksson, J. M., Andersen, L. M., & Bejerot, S. (2013). RAADS-14 screen: validity of a screening tool for autism spectrum disorder in an adult psychiatric population. *Molecular Autism*, *4*(1), 49.

Erlangsen, A., Jacobsen, A. L., Ranning, A., Delamare, A. L., Nordentoft, M., & Frisch, M. (2023). Transgender identity and suicide attempts and mortality in Denmark. *JAMA*, *329*(24), 2145–2153.

Evans, S. F., Raymond, S., Sethuram, S., Barrett, E. S., Bush, N. R., Nguyen, R., Sathyanarayana, S., & Swan, S. H. (2021). Associations between prenatal phthalate exposure and sex-typed play behavior in preschool age boys and girls. *Environmental Research*, *192*, 110264.

Fast, A. A., & Olson, K. R. (2018). Gender development in transgender preschool children. *Child Development*, *89*(2), 620–637.

Fernández, R., Esteva, I., Gómez-Gil, E., Rumbo, T., Almaraz, M. C., Roda, E., Haro-Mora, J. J., Guillamón, A., & Pásaro, E. (2014). The (CA)n polymorphism of ERβ gene is associated with FtM transsexualism. *The Journal of Sexual Medicine, 11*(3), 720–728.

Fernández, R., Guillamon, A., Cortés-Cortés, J., Gómez-Gil, E., Jácome, A., Esteva, I., Almaraz, M., Mora, M., Aranda, G., & Pásaro, E. (2018). Molecular basis of gender dysphoria: androgen and estrogen receptor interaction. *Psychoneuroendocrinology, 98*, 161–167.

Fischbach, A. L., Hindenach, A., van der Miesen, A. I. R., Yang, J. S., Buckley, O. J., Song, M., Campos, L., & Strang, J. F. (2024). Autistic and non-autistic transgender youth are similar in gender development and sexuality phenotypes. *The British Journal of Developmental Psychology*, 10.1111/bjdp.12486. Advance online publication.

Foreman, M., Hare, L., York, K., Balakrishnan, K., Sánchez, F. J., Harte, F., Erasmus, J., Vilain, E., & Harley, V. R. (2019). Genetic link between gender dysphoria and sex hormone signaling. *The Journal of Clinical Endocrinology and Metabolism, 104*(2), 390–396.

Fortunato, A., Giovanardi, G., Innocenzi, E., Mirabella, M., Caviglia, G., Lingiardi, V., & Speranza, A. M. (2022). Is it autism? A critical commentary on the co-occurrence of gender dysphoria and autism spectrum disorder. *Journal of Homosexuality*, *69*(7), 1204–1221.

Gavazzi, G., Fisher, A. D., Orsolini, S., Bianchi, A., Romani, A., Giganti, F., Giovannelli, F., Ristori, J., Mazzoli, F., Maggi, M., Viggiano, M. P., & Mascalchi, M. (2022). The fMRI correlates of visuo-spatial abilities: sex differences and gender dysphoria. *Brain Imaging and Behavior*, *16*(2), 955–964.

Goh, T. J., Chua, D. K., Vasudevan, K., & Fung, D. S. S. (2024). Gender dysphoria in children and adolescents: a retrospective analysis of cases in Singapore. *Annals of the Academy of Medicine, Singapore*, *53*(11), 647–656.

Golombok, S., & Rust, J. (1993a). The measurement of gender role behaviour in pre-school children: a research note. *Journal of Child Psychology and Psychiatry and Allied Disciplines*, *34*(5), 805–811.

Golombok, S., & Rust, J. (1993b). The Pre-School Activities Inventory: a standardized assessment of gender role in children. *Psychological Assessment*, *5*(2), 131.

Goodman R. (1997). The Strengths and Difficulties Questionnaire: a research note. *Journal of Child Psychology and Psychiatry and Allied Disciplines*, *38*(5), 581–586.

Goodman, M., Adams, N., Corneil, T., Kreukels, B., Motmans, J., & Coleman, E. (2019). Size and distribution of transgender and gender nonconforming populations: a narrative review. *Endocrinology and Metabolism Clinics of North America*, *48*(2), 303–321.

Gülgöz, S., Glazier, J. J., Enright, E. A., Alonso, D. J., Durwood, L. J., Fast, A. A., Lowe, R., Ji, C., Heer, J., Martin, C. L., & Olson, K. R. (2019). Similarity in transgender and cisgender children's gender development. *Proceedings of the National Academy of Sciences of the United States of America*, *116*(49), 24480–24485.

Hendriks, O., Wei, Y., Warrier, V., & Richards, G. (2022). Autistic traits, empathizing-systemizing, and gender diversity. *Archives of Sexual Behavior*, *51*(4), 2077–2089.

Henningsson, S., Westberg, L., Nilsson, S., Lundström, B., Ekselius, L., Bodlund, O., Lindström, E., Hellstrand, M., Rosmond, R., Eriksson, E., & Landén, M. (2005). Sex steroid-related genes and male-to-female transsexualism. *Psychoneuroendocrinology*, *30*(7), 657–664.

Hines M. (2020). Human gender development. *Neuroscience and Biobehavioral Reviews*, *118*, 89–96.

Hines, M., Ahmed, S. F., & Hughes, I. A. (2003). Psychological outcomes and gender-related development in complete androgen insensitivity syndrome. *Archives of Sexual Behavior*, *32*(2), 93–101.

Hines, M., Golombok, S., Rust, J., Johnston, K. J., Golding, J., & Avon Longitudinal Study of Parents and Children Study Team (2002). Testosterone during pregnancy and gender role behavior of preschool children: a longitudinal, population study. *Child Development*, *73*(6), 1678–1687.

Hlisníková, H., Petrovičová, I., Kolena, B., Šidlovská, M., & Sirotkin, A. (2020). Effects and mechanisms of phthalates' action on reproductive processes and reproductive health: a literature review. *International Journal of Environmental Research and Public Health*, *17*(18), 6811.

Hoekzema, E., Schagen, S. E., Kreukels, B. P., Veltman, D. J., Cohen-Kettenis, P. T., Delemarre-van de Waal, H., & Bakker, J. (2015). Regional volumes and spatial volumetric distribution of gray matter in the gender dysphoric brain. *Psychoneuroendocrinology*, *55*, 59–71.

Hood E. (2005). Are EDCs blurring issues of gender?. *Environmental Health Perspectives*, *113*(10), A670–A677.

Huisman, B., Noens, I., Steensma, T. D., Kreukels, B. P. C., & van der Miesen, A. I. R. (2024). Autism traits in transgender and gender-diverse adults seeking gender-affirming medical treatment. *International Journal of Transgender Health,* 1–17.

Iervolino, A. C., Hines, M., Golombok, S. E., Rust, J., & Plomin, R. (2005). Genetic and environmental influences on sex-typed behavior during the preschool years. *Child Development*, *76*(4), 826–840.

Indremo, M., White, R., Frisell, T., Cnattingius, S., Skalkidou, A., Isaksson, J., & Papadopoulos, F. C. (2021). Validity of the gender dysphoria diagnosis and incidence trends in Sweden: a nationwide register study. *Scientific Reports*, *11*(1), 16168.

Janssen, A., Huang, H., & Duncan, C. (2016). Gender variance among youth with autism spectrum disorders: a retrospective chart review. *Transgender Health*, *1*(1), 63–68.

Jarvis, S. W., Fraser, L. K., Langton, T., Hewitt, C. E., & Doran, T. (2025). Epidemiology of gender dysphoria and gender incongruence in children and young people attending primary care practices in England: retrospective cohort study. *Archives of Disease in Childhood*, archdischild-2024-327992. Advance online publication.

Joel D. (2021). Beyond the binary: rethinking sex and the brain. *Neuroscience and Biobehavioral Reviews*, *122*, 165–175.

Johnson, A. H. (2016). Transnormativity: a new concept and its validation through documentary film about transgender men. *Sociological Inquiry*, *86*(4), 465–491.

Jones, R. M., Wheelwright, S., Farrell, K., Martin, E., Green, R., Di Ceglie, D., & Baron-Cohen, S. (2012). Brief report: female-to-male transsexual people and autistic traits. *Journal of Autism and Developmental Disorders*, *42*(2), 301–306.

Jürgensen, M., Hiort, O., Holterhus, P. M., & Thyen, U. (2007). Gender role behavior in children with XY karyotype and disorders of sex development. *Hormones and Behavior*, *51*(3), 443–453.

Kahn, L. G., Philippat, C., Nakayama, S. F., Slama, R., & Trasande, L. (2020). Endocrine-disrupting chemicals: implications for human health. *The Lancet. Diabetes & Endocrinology*, *8*(8), 703–718.

Kallitsounaki, A., & Williams, D. M. (2022). Implicit and explicit gender-related cognition, gender dysphoria, autistic-like traits, and mentalizing: differences between autistic and non-autistic cisgender and transgender adults. *Archives of Sexual Behavior*, *51*(7), 3583–3600.
Kallitsounaki, A., & Williams, D. M. (2023). Autism spectrum disorder and gender dysphoria/incongruence. A systematic literature review and meta-analysis. *Journal of Autism and Developmental Disorders*, *53*(8), 3103–3117.
Kaltiala, R., Bergman, H., Carmichael, P., de Graaf, N. M., Egebjerg Rischel, K., Frisén, L., Schorkopf, M., Suomalainen, L., & Waehre, A. (2020). Time trends in referrals to child and adolescent gender identity services: a study in four Nordic countries and in the UK. *Nordic Journal of Psychiatry*, *74*(1), 40–44.
Kaltiala, R., Holttinen, T., & Tuisku, K. (2023). Have the psychiatric needs of people seeking gender reassignment changed as their numbers increase? A register study in Finland. *European Psychiatry: The Journal of the Association of European Psychiatrists*, *66*(1), e93.
Kennis, M., Staicu, R., Dewitte, M., T'Sjoen, G., Sack, A. T., & Duecker, F. (2024). Heed lessons from past studies involving transgender people: first, do no harm. *Nature*, *629*(8014), 998–1000.
Klinger, D., Oehlke, S. M., Riedl, S., Eschbaum, K., Zesch, H. E., Karwautz, A., Plener, P. L., & Kothgassner, O. D. (2024). Mental health of non-binary youth: a systematic review and meta-analysis. *Child and Adolescent Psychiatry and Mental Health*, *18*(1), 126.
Knafo, A., Iervolino, A. C., & Plomin, R. (2005). Masculine girls and feminine boys: genetic and environmental contributions to atypical gender development in early childhood. *Journal of Personality and Social Psychology*, *88*(2), 400–412.
Knickmeyer, R. C., Wheelwright, S., Taylor, K., Raggatt, P., Hackett, G., & Baron-Cohen, S. (2005). Gender-typed play and amniotic testosterone. *Developmental Psychology*, *41*(3), 517–528.
Koehler, A., Richter-Appelt, H., Cerwenka, S., Kreukels, B. P., Watzlawik, M., Cohen-Kettenis, P. T., de Cuypere, G., Haraldsen, I. R. H., & Nieder, T. O. (2017). Recalled gender-related play behavior and peer-group preferences in childhood and adolescence among adults applying for gender-affirming treatment. *Sexual and Relationship Therapy*, *32*(2), 210–226.
Kozee, H. B., Tylka, T. L., & Bauerband, L. A. (2012). Measuring transgender individuals' comfort with gender identity and appearance: development and validation of the Transgender Congruence Scale. *Psychology of Women Quarterly*, *36*(2), 179–196.
Kung K. T. F. (2020). Autistic traits, systemising, empathising, and theory of mind in transgender and non-binary adults. *Molecular Autism*, *11*(1), 73.
Kung, K. T., Constantinescu, M., Browne, W. V., Noorderhaven, R. M., & Hines, M. (2016). No relationship between early postnatal testosterone concentrations and autistic traits in 18 to 30-month-old children. *Molecular Autism*, *7*, 15.
Kung, K. T. F., Louie, K., Spencer, D., & Hines, M. (2024). Prenatal androgen exposure and sex-typical play behaviour: a meta-analysis of classic congenital adrenal hyperplasia studies. *Neuroscience and Biobehavioral Reviews*, *159*, 105616.
Kuper, L. E., Wright, L., & Mustanski, B. (2018). Gender identity development among transgender and gender nonconforming emerging adults: an intersectional approach. *International Journal of Transgenderism*, *19*(4), 436–455.

La Merrill, M. A., Vandenberg, L. N., Smith, M. T., Goodson, W., Browne, P., Patisaul, H. B., Guyton, K. Z., Kortenkamp, A., Cogliano, V. J., Woodruff, T. J., Rieswijk, L., Sone, H., Korach, K. S., Gore, A. C., Zeise, L., & Zoeller, R. T. (2020). Consensus on the key characteristics of endocrine-disrupting chemicals as a basis for hazard identification. *Nature Reviews. Endocrinology*, *16*(1), 45–57.

Lamb, M. E., Easterbrooks, M. A., & Holden, G. W. (1980). Reinforcement and punishment among preschoolers: characteristics, effects, and correlates. *Child Development*, 1230–1236.

Lamminmäki, A., Hines, M., Kuiri-Hänninen, T., Kilpeläinen, L., Dunkel, L., & Sankilampi, U. (2012). Testosterone measured in infancy predicts subsequent sex-typed behavior in boys and in girls. *Hormones and Behavior*, *61*(4), 611–616.

Langlois, J. H., & Downs, A. C. (1980). Mothers, fathers, and peers as socialization agents of sex-typed play behaviors in young children. *Child Development*, 1237–1247.

Lev, A. I. (2004). Transgender emergence: a developmental process. In T. Trepper (Ed.), *Transgender emergence: therapeutic guidelines for working with gender-variant people and their families* (pp. 229–269). The Haworth Clinical Practice Press.

Levitt, H. M., & Ippolito, M. R. (2013). Being transgender: navigating minority stressors and developing authentic self-presentation. *Psychology of Women Quarterly*, *38*(1), 46–64.

Li, G., Kung, K. T., & Hines, M. (2017). Childhood gender-typed behavior and adolescent sexual orientation: a longitudinal population-based study. *Developmental Psychology*, *53*(4), 764–777.

Lian, Q., Li, R., Liu, Z., Li, X., Su, Q., & Zheng, D. (2022). Associations of nonconforming gender expression and gender identity with bullying victimization: an analysis of the 2017 youth risk behavior survey. *BMC Public Health*, *22*(1), 650.

Liu, J., Gao, D., Wang, H., Li, Y., Chen, M., Ma, Q., Wang, X., Cui, M., Chen, L., Zhang, Y., Guo, T., Yuan, W., Ma, T., Jiang, J., Dong, Y., Zou, Z., & Ma, J. (2023). Long-term exposure to exogenous phthalate, masculinity and femininity trait, and gender identity in children: a Chinese 3-year longitudinal cohort study. *Environmental Health: A Global Access Science Source*, *22*(1), 81.

Lord, C., Brugha, T. S., Charman, T., Cusack, J., Dumas, G., Frazier, T., Jones, E. J. H., Jones, R. M., Pickles, A., State, M. W., Taylor, J. L., & Veenstra-VanderWeele, J. (2020). Autism spectrum disorder. *Nature Reviews. Disease Primers*, *6*(1), 5.

Lowry, R., Johns, M. M., Gordon, A. R., Austin, S. B., Robin, L. E., & Kann, L. K. (2018). Nonconforming gender expression and associated mental distress and substance use among high school students. *JAMA Pediatrics*, *172*(11), 1020–1028.

Mahfouda, S., Panos, C., Whitehouse, A. J. O., Thomas, C. S., Maybery, M., Strauss, P., Zepf, F. D., O'Donovan, A., van Hall, H. W., Saunders, L. A., Moore, J. K., & Lin, A. (2019). Mental health correlates of autism spectrum disorder in gender diverse young people: evidence from a specialised child and adolescent gender clinic in Australia. *Journal of Clinical Medicine*, *8*(10), 1503.

Manzouri, A., & Savic, I. (2019). Possible neurobiological underpinnings of homosexuality and gender dysphoria. *Cerebral Cortex (New York, N.Y.: 1991)*, *29*(5), 2084–2101.

Marino, J. L., Lin, A., Davies, C., Kang, M., Bista, S., & Skinner, S. R. (2023). Childhood and adolescence gender role nonconformity and gender and sexuality diversity in young adulthood. *JAMA Pediatrics*, *177*(11), 1176–1186.

Martin, C. L., Ruble, D. N., & Szkrybalo, J. (2002). Cognitive theories of early gender development. *Psychological Bulletin*, *128*(6), 903–933.

Matsuno, E., & Budge, S. L. (2017). Non-binary/genderqueer identities: a critical review of the literature. *Current Sexual Health Reports*, *9*, 116–120.

McGuire, J. K., & Morrow, Q. J. (2020). Pathways of gender development. In M. Forcier, G. van Schalkwyk, & J. L. Turban (Eds.), *Pediatric gender identity: gender-affirming care for transgender & gender diverse youth* (pp. 33–46). Springer.

McLean, K. C., Shucard, H., & Syed, M. (2017). Applying the master narrative framework to gender identity development in emerging adulthood. *Emerging Adulthood*, *5*(2), 93–105.

Metcalfe, C. D., Bayen, S., Desrosiers, M., Muñoz, G., Sauvé, S., & Yargeau, V. (2022). An introduction to the sources, fate, occurrence and effects of endocrine disrupting chemicals released into the environment. *Environmental Research*, *207*, 112658.

Meyer I. H. (2003). Prejudice, social stress, and mental health in lesbian, gay, and bisexual populations: conceptual issues and research evidence. *Psychological Bulletin*, *129*(5), 674–697.

Meyer-Bahlburg, H. F., Dolezal, C., Baker, S. W., Ehrhardt, A. A., & New, M. I. (2006). Gender development in women with congenital adrenal hyperplasia as a function of disorder severity. *Archives of Sexual Behavior*, *35*(6), 667–684.

Mezza, F., Mezzalira, S., Pizzo, R., Maldonato, N. M., Bochicchio, V., & Scandurra, C. (2024). Minority stress and mental health in European transgender and gender diverse people: a systematic review of quantitative studies. *Clinical Psychology Review*, *107*, 102358.

Miller, L. R., & Grollman, E. A. (2015). The social costs of gender nonconformity for transgender adults: implications for discrimination and health. *Sociological Forum (Randolph, N.J.)*, *30*(3), 809–831.

Mischel, W. (1966). A social-learning view of sex differences in behavior. In E. Maccoby (Ed.), *The development of sex differences* (pp. 56–81). Stanford University Press.

Modica, R., Benevento, E., & Colao, A. (2023). Endocrine-disrupting chemicals (EDCs) and cancer: new perspectives on an old relationship. *Journal of Endocrinological Investigation*, *46*(4), 667–677.

Narita, Z., DeVylder, J., Yamasaki, S., Ando, S., Endo, K., Miyashita, M., Yamaguchi, S., Usami, S., Stanyon, D., Knowles, G., Hiraiwa-Hasegawa, M., Furukawa, T. A., Kasai, K., & Nishida, A. (2024). Uncovering associations between gender nonconformity, psychosocial factors, and mental health in adolescents: a birth cohort study. *Psychological Medicine*, *54*(5), 921–930.

Newhook, J. T., Pyne, J., Winters, K., Feder, S., Holmes, C., Tosh, J., Sinnott, M., Jamieson, A., & Pickett, S. (2018). A critical commentary on follow-up studies and “desistance” theories about transgender and gender-nonconforming children. *International Journal of Transgenderism, 19*(2), 212–224.

Nobili, A., Glazebrook, C., Bouman, W. P., Glidden, D., Baron-Cohen, S., Allison, C., Smith, P., & Arcelus, J. (2018). Autistic traits in treatment-seeking transgender adults. *Journal of Autism and Developmental Disorders*, *48*(12), 3984–3994.

Nordenström, A., Servin, A., Bohlin, G., Larsson, A., & Wedell, A. (2002). Sex-typed toy play behavior correlates with the degree of prenatal androgen exposure assessed by CYP21 genotype in girls with congenital adrenal hyperplasia. *The Journal of Clinical Endocrinology and Metabolism*, *87*(11), 5119–5124.

Nyquist, C. B., Torgersen, L., David, L. W., Diseth, T. H., Magnus, P., Biele, G. P. E., & Waehre, A. (2025). Population-adjusted numbers, demographics and mental health among children and adolescents referred to the Norwegian National Center for Gender Incongruence over two decades. *European Child & Adolescent Psychiatry*, *34*(2), 647–657.

Oginni, O. A., Robinson, E. J., Jones, A., Rahman, Q., & Rimes, K. A. (2019). Mediators of increased self-harm and suicidal ideation in sexual minority youth: a longitudinal study. *Psychological Medicine*, *49*(15), 2524–2532.

Olson, K. R., Key, A. C., & Eaton, N. R. (2015). Gender cognition in transgender children. *Psychological Science*, *26*(4), 467–474.

Özel, F., Indremo, M., Karamanis, G., Elofsson, U., Beckman, U., Fazekas, A., Frisén, L., Isaksson, M., Sandström, L., Thelin, N., Tivesten, Å., Wahlberg, J., Skalkidou, A., Bodlund, O., & Papadopoulos, F. C. (2023). Exploring gender dysphoria and related outcomes in a prospective cohort study: protocol for the Swedish Gender Dysphoria Study (SKDS). *BMJ Open*, *13*(4), e066571.

Parker, C., Scott, S., & Geddes, A. (2019). Snowball sampling. In P. Atkinson, S. Delamont, A. Cernat, J. W. Sakshaug, & R. A. Williams (Eds.), *SAGE research methods foundations.* SAGE.

Pasterski, V., Geffner, M. E., Brain, C., Hindmarsh, P., Brook, C., & Hines, M. (2011). Prenatal hormones and childhood sex segregation: playmate and play style preferences in girls with congenital adrenal hyperplasia. *Hormones and Behavior*, *59*(4), 549–555.

Pasterski, V., Zucker, K. J., Hindmarsh, P. C., Hughes, I. A., Acerini, C., Spencer, D., Neufeld, S., & Hines, M. (2015). Increased cross-gender identification independent of gender role behavior in girls with congenital adrenal hyperplasia: results from a standardized assessment of 4- to 11-year-old children. *Archives of Sexual Behavior*, *44*(5), 1363–1375.

Pellicane, M. J., & Ciesla, J. A. (2022). Associations between minority stress, depression, and suicidal ideation and attempts in transgender and gender diverse (TGD) individuals: systematic review and meta-analysis. *Clinical Psychology Review*, *91*, 102113.

Percy, Z., Xu, Y., Sucharew, H., Khoury, J. C., Calafat, A. M., Braun, J. M., Lanphear, B. P., Chen, A., & Yolton, K. (2016). Gestational exposure to phthalates and gender-related play behaviors in 8-year-old children: an observational study. *Environmental Health: A Global Access Science Source*, *15*(1), 87.

Pullen Sansfaçon, A., Medico, D., Suerich-Gulick, F., & Temple Newhook, J. (2020). "I knew that I wasn't cis, I knew that, but I didn't know exactly": gender identity development, expression and affirmation in youth who access gender affirming medical care. *International Journal of Transgender Health*, *21*(3), 307–320.

Ramirez, K., Fernández, R., Collet, S., Kiyar, M., Delgado-Zayas, E., Gómez-Gil, E., van den Eynde, T., T'Sjoen, G., Guillamon, A., Mueller, S. C., & Pásaro, E. (2021). Epigenetics is implicated in the basis of gender incongruence: an epigenome-wide association analysis. *Frontiers in Neuroscience*, *15*, 701017.

Ramos, N., & Marr, M. C. (2023). Traumatic stress and resilience among transgender and gender diverse youth. *Child and Adolescent Psychiatric Clinics of North America*, *32*(4), 667–682.

Reisner, S. L., Poteat, T., Keatley, J., Cabral, M., Mothopeng, T., Dunham, E., Holland, C. E., Max, R., & Baral, S. D. (2016). Global health burden and needs of transgender populations: a review. *Lancet (London, England)*, *388*(10042), 412–436.

Ridley, R. (2019). Some difficulties behind the concept of the 'extreme male brain' in autism research. A theoretical review. *Research in Autism Spectrum Disorders*, *57*, 19–27.

Riggs, D. W. (2019). *Working with transgender young people and their families: a critical developmental approach*. Springer International Publishing.

Roshan, G. M., Talaei, A., Sadr, M., Arezoomandan, S., Kazemi, S., & Khorashad, B. S. (2019). Recalled pre-school activities among adults with gender dysphoria who seek gender confirming treatment - an Iranian study. *Asian Journal of Psychiatry*, *42*, 57–61.

Sasaki, S., Ozaki, K., Yamagata, S., Takahashi, Y., Shikishima, C., Kornacki, T., Nonaka, K., & Ando, J. (2016). Genetic and environmental influences on traits of gender identity disorder: a study of Japanese twins across developmental stages. *Archives of Sexual Behavior*, *45*(7), 1681–1695.

Schiltz, H. K., McVey, A. J., van Dyk, I. S., Adler, E. J., & Van Hecke, A. V. (2023). Brief report: links between nonconformity to gender norms, autistic features, and internalizing symptoms in a non-clinical college sample. *Journal of Autism and Developmental Disorders*, *53*(4), 1717–1725.

Servin, A., Nordenström, A., Larsson, A., & Bohlin, G. (2003). Prenatal androgens and gender-typed behavior: a study of girls with mild and severe forms of congenital adrenal hyperplasia. *Developmental Psychology*, *39*(3), 440–450.

Shumer, D. E., Roberts, A. L., Reisner, S. L., Lyall, K., & Austin, S. B. (2015). Brief report: autistic traits in mothers and children associated with child's gender nonconformity. *Journal of Autism and Developmental Disorders*, *45*(5), 1489–1494.

Skagerberg, E., Di Ceglie, D., & Carmichael, P. (2015). Brief report: autistic features in children and adolescents with gender dysphoria. *Journal of Autism and Developmental Disorders*, *45*(8), 2628–2632.

Smith, E. S., Junger, J., Derntl, B., & Habel, U. (2015). The transsexual brain - a review of findings on the neural basis of transsexualism. *Neuroscience and Biobehavioral Reviews*, *59*, 251–266.

Socialstyrelsen. (2020). *Utvecklingen av diagnosen könsdysfori.* https://www.socialstyrelsen.se/globalassets/sharepoint-dokument/artikelkatalog/ovrigt/2020-2-6600.pdf

Socialstyrelsen. (2025). *Stärkt uppföljning av vård vid könsdysfori.* https://www.socialstyrelsen.se/globalassets/sharepoint-dokument/artikelkatalog/ovrigt/2025-2-9418.pdf

Speechley, M., Stuart, J., & Modecki, K. L. (2024). Diverse gender identity development: a qualitative synthesis and development of a new contemporary framework. *Sex Roles*, *90*(1), 1–18.

Spivey, L. A., Huebner, D. M., & Diamond, L. M. (2018). Parent responses to childhood gender nonconformity: effects of parent and child characteristics. *Psychology of Sexual Orientation and Gender Diversity, 5*(3), 360–370.

Steensma, T. D., Biemond, R., de Boer, F., & Cohen-Kettenis, P. T. (2011). Desisting and persisting gender dysphoria after childhood: a qualitative follow-up study. *Clinical Child Psychology and Psychiatry*, *16*(4), 499–516.

Steensma, T. D., Cohen-Kettenis, P. T., & Zucker, K. J. (2018). Evidence for a change in the sex ratio of children referred for gender dysphoria: data from the center of expertise on gender dysphoria in Amsterdam (1988-2016). *Journal of Sex & Marital Therapy*, *44*(7), 713–715.

Steensma, T. D., McGuire, J. K., Kreukels, B. P., Beekman, A. J., & Cohen-Kettenis, P. T. (2013b). Factors associated with desistence and persistence of childhood gender dysphoria: a quantitative follow-up study. *Journal of the American Academy of Child and Adolescent Psychiatry*, *52*(6), 582–590.

Steensma, T. D., van der Ende, J., Verhulst, F. C., & Cohen-Kettenis, P. T. (2013a). Gender variance in childhood and sexual orientation in adulthood: a prospective study. *The Journal of Sexual Medicine*, *10*(11), 2723–2733.

Strang, J. F., Anthony, L. G., Song, A., Lai, M. C., Knauss, M., Sadikova, E., Graham, E., Zaks, Z., Wimms, H., Willing, L., Call, D., Mancilla, M., Shakin, S., Vilain, E., Kim, D. Y., Maisashvili, T., Khawaja, A., & Kenworthy, L. (2023). In addition to stigma: cognitive and autism-related predictors of mental health in transgender adolescents. *Journal of Clinical Child and Adolescent Psychology: The Official Journal for the Society of Clinical Child and Adolescent Psychology, American Psychological Association, Division 53*, *52*(2), 212–229.

Strang, J. F., Kenworthy, L., Dominska, A., Sokoloff, J., Kenealy, L. E., Berl, M., Walsh, K., Menvielle, E., Slesaransky-Poe, G., Kim, K. E., Luong-Tran, C., Meagher, H., & Wallace, G. L. (2014). Increased gender variance in autism spectrum disorders and attention deficit hyperactivity disorder. *Archives of Sexual Behavior*, *43*(8), 1525–1533.

Strang, J. F., Meagher, H., Kenworthy, L., de Vries, A. L. C., Menvielle, E., Leibowitz, S., Janssen, A., Cohen-Kettenis, P., Shumer, D. E., Edwards-Leeper, L., Pleak, R. R., Spack, N., Karasic, D. H., Schreier, H., Balleur, A., Tishelman, A., Ehrensaft, D., Rodnan, L., Kuschner, E. S., … Anthony, L. G. (2018). Initial clinical guidelines for co-occurring autism spectrum disorder and gender dysphoria or incongruence in adolescents. *Journal of Clinical Child and Adolescent Psychology: The Official Journal for the Society of Clinical Child and Adolescent Psychology, American Psychological Association, Division 53*, *47*(1), 105–115.

Strauss, P., Cook, A., Watson, V., Winter, S., Whitehouse, A., Albrecht, N., Wright Toussaint, D., & Lin, A. (2021). Mental health difficulties among trans and gender diverse young people with an autism spectrum disorder (ASD): findings from Trans Pathways. *Journal of Psychiatric Research*, *137*, 360–367.

Sumia, M., Lindberg, N., Työläjärvi, M., & Kaltiala-Heino, R. (2017). Current and recalled childhood gender identity in community youth in comparison to referred adolescents seeking sex reassignment. *Journal of Adolescence*, *56*, 34–39.

Swan, S. H., Liu, F., Hines, M., Kruse, R. L., Wang, C., Redmon, J. B., Sparks, A., & Weiss, B. (2010). Prenatal phthalate exposure and reduced masculine play in boys. *International Journal of Andrology*, *33*(2), 259–269.

Taboas, A., Doepke, K., & Zimmerman, C. (2023). Preferences for identity-first versus person-first language in a US sample of autism stakeholders. *Autism: The International Journal of Research and Practice*, *27*(2), 565–570.

Tanner, E. M., Bornehag, C. G., & Gennings, C. (2019). Repeated holdout validation for weighted quantile sum regression. *MethodsX*, *6*, 2855–2860.

Thorne, N., Yip, A. K., Bouman, W. P., Marshall, E., & Arcelus, J. (2019). The terminology of identities between, outside and beyond the gender binary - a systematic review. *International Journal of Transgenderism*, *20*(2-3), 138–154.

Tollit, M. A., Maloof, T., Hoq, M., Haebich, K., Pace, C. C., Rodriguez, Z. M., Sial, M., Payne, J. M., & Pang, K. (2024). A comparison of gender diversity in transgender young people with and without autistic traits from the Trans 20 cohort study. *The Lancet Regional Health. Western Pacific*, *47*, 101084.

Troisi, R., Palmer, J. R., Hatch, E. E., Strohsnitter, W. C., Huo, D., Hyer, M., Fredriksen-Goldsen, K. I., Hoover, R., & Titus, L. (2020). Gender identity and sexual orientation identity in women and men prenatally exposed to diethylstilbestrol. *Archives of Sexual Behavior*, *49*(2), 447–454.

Turban, J. L., & van Schalkwyk, G. I. (2018). "Gender dysphoria" and autism spectrum disorder: Is the link real?. *Journal of the American Academy of Child and Adolescent Psychiatry*, *57*(1), 8–9.e2.

Üstay, Ö., Elbasan, O., Erel, P., Bulut, N. S., & Yorguner, N. (2024). Endocrine-disrupting effects of bisphenol-A, thiamethoxam, and fipronil in hormone-naïve transmen compared to cis-women. *Hormones (Athens, Greece)*, *23*(3), 375–383.

van Beijsterveldt, C. E., Hudziak, J. J., & Boomsma, D. I. (2006). Genetic and environmental influences on cross-gender behavior and relation to behavior problems: a study of Dutch twins at ages 7 and 10 years. *Archives of Sexual Behavior*, *35*(6), 647–658.

van Beusekom, G., Collier, K. L., Bos, H. M. W., Sandfort, T. G. M., & Overbeek, G. (2020). Gender nonconformity and peer victimization: sex and sexual attraction differences by age. *Journal of Sex Research*, *57*(2), 234–246.

van de Beek, C., van Goozen, S. H., Buitelaar, J. K., & Cohen-Kettenis, P. T. (2009). Prenatal sex hormones (maternal and amniotic fluid) and gender-related play behavior in 13-month-old infants. *Archives of Sexual Behavior*, *38*(1), 6–15.

van der Miesen, A. I. R., de Vries, A. L. C., Steensma, T. D., & Hartman, C. A. (2018a). Autistic symptoms in children and adolescents with gender dysphoria. *Journal of Autism and Developmental Disorders*, *48*(5), 1537–1548.

van der Miesen, A. I. R., Hurley, H., Bal, A. M., & de Vries, A. L. C. (2018b). Prevalence of the wish to be of the opposite gender in adolescents and adults with autism spectrum disorder. *Archives of Sexual Behavior*, *47*(8), 2307–2317.

van der Miesen, A. I., Hurley, H., & de Vries, A. L. (2016). Gender dysphoria and autism spectrum disorder: a narrative review. *International Review of Psychiatry (Abingdon, England)*, *28*(1), 70–80.

van der Miesen, A. I. R., Nabbijohn, A. N., Santarossa, A., & VanderLaan, D. P. (2018c). Behavioral and emotional problems in gender-nonconforming children: a canadian community-based study. *Journal of the American Academy of Child and Adolescent Psychiatry*, *57*(7), 491–499.

Vreugdenhil, H. J., Slijper, F. M., Mulder, P. G., & Weisglas-Kuperus, N. (2002). Effects of perinatal exposure to PCBs and dioxins on play behavior in Dutch children at school age. *Environmental Health Perspectives*, *110*(10), A593–A598.

Wallien, M. S., & Cohen-Kettenis, P. T. (2008). Psychosexual outcome of gender-dysphoric children. *Journal of the American Academy of Child and Adolescent Psychiatry*, *47*(12), 1413–1423.

Warrier, V., Greenberg, D. M., Weir, E., Buckingham, C., Smith, P., Lai, M. C., Allison, C., & Baron-Cohen, S. (2020). Elevated rates of autism, other neurodevelopmental and psychiatric diagnoses, and autistic traits in transgender and gender-diverse individuals. *Nature Communications*, *11*(1), 3959.

Wattel, L. L., Walsh, R. J., & Krabbendam, L. (2024). Theories on the link between autism spectrum conditions and trans gender modality: a systematic review. *Review Journal of Autism and Developmental Disorders*, *11*(2), 275–295.

Whitehouse, A. J., Mattes, E., Maybery, M. T., Dissanayake, C., Sawyer, M., Jones, R. M., Pennell, C. E., Keelan, J. A., & Hickey, M. (2012). Perinatal testosterone exposure and autistic-like traits in the general population: a longitudinal pregnancy-cohort study. *Journal of Neurodevelopmental Disorders*, *4*(1), 25.

Wiepjes, C. M., Nota, N. M., de Blok, C. J. M., Klaver, M., de Vries, A. L. C., Wensing-Kruger, S. A., de Jongh, R. T., Bouman, M. B., Steensma, T. D., Cohen-Kettenis, P., Gooren, L. J. G., Kreukels, B. P. C., & den Heijer, M. (2018). The Amsterdam Cohort of Gender Dysphoria Study (1972-2015): trends in prevalence, treatment, and regrets. *The Journal of Sexual Medicine*, *15*(4), 582–590.

Winneke, G., Ranft, U., Wittsiepe, J., Kasper-Sonnenberg, M., Fürst, P., Krämer, U., Seitner, G., & Wilhelm, M. (2014). Behavioral sexual dimorphism in school-age children and early developmental exposure to dioxins and PCBs: a follow-up study of the Duisburg cohort. *Environmental Health Perspectives*, *122*(3), 292–298.

Wirtz, A. L., Poteat, T. C., Malik, M., & Glass, N. (2020). Gender-based violence against transgender people in the United States: a call for research and programming. *Trauma, Violence & Abuse*, *21*(2), 227–241.

Witcomb, G. L., Bouman, W. P., Claes, L., Brewin, N., Crawford, J. R., & Arcelus, J. (2018). Levels of depression in transgender people and its predictors: results of a large matched control study with transgender people accessing clinical services. *Journal of Affective Disorders*, *235*, 308–315.

World Health Organization. (2002). *Global assessment of the state-of-the-science of endocrine disruptors.* https://iris.who.int/bitstream/handle/10665/67357/WHO_PCS_EDC_02.2.pdf?sequence=1

World Health Organization. (2019). *International classification of diseases for mortality and morbidity statistics* (11th ed.). World Health Organization.

World Health Organization. (2025). *Gender and health.* https://www.who.int/health-topics/gender#tab=tab_1

Xerxa, Y., White, T., Busa, S., Trasande, L., Hillegers, M. H. J., Jaddoe, V. W., Castellanos, F. X., & Ghassabian, A. (2023). Gender diversity and brain morphology among adolescents. *JAMA Network Open*, *6*(5), e2313139.

Xu, Y., Feng, J., & Rahman, Q. (2024). Gender nonconformity and common mental health problems: a meta-analysis. *Clinical Psychology Review*, *114*, 102500.

Zeidan, J., Fombonne, E., Scorah, J., Ibrahim, A., Durkin, M. S., Saxena, S., Yusuf, A., Shih, A., & Elsabbagh, M. (2022). Global prevalence of autism: a systematic review update. *Autism Research: Official Journal of the International Society for Autism Research*, *15*(5), 778–790.

Zhang, Q., Goodman, M., Adams, N., Corneil, T., Hashemi, L., Kreukels, B., Motmans, J., Snyder, R., & Coleman, E. (2020). Epidemiological considerations in transgender health: a systematic review with focus on higher quality data. *International Journal of Transgender Health*, *21*(2), 125–137.

Zhang, Q., Rechler, W., Bradlyn, A., Flanders, W. D., Getahun, D., Lash, T. L., McCracken, C., Nash, R., Panagiotakopoulos, L., Roblin, D., Sandberg, D. E., Silverberg, M. J., Tangpricha, V., Vupputuri, S., & Goodman, M. (2021). Changes in size and demographic composition of transgender and gender nonbinary population receiving care at integrated health systems. *Endocrine Practice: Official Journal of the American College of Endocrinology and the American Association of Clinical Endocrinologists*, *27*(5), 390–395.

Zhao, M., Xiao, D., Wang, W., Wu, R., Dewaele, A., Zhang, W., Buysse, A., Song, C., Guo, L., & Lu, C. (2021). Association of sexual minority status, gender nonconformity with childhood victimization and adulthood depressive symptoms: a path analysis. *Child Abuse & Neglect*, *111*, 104822.

Zhu, Y. D., Han, X., Wang, X. Q., Ge, T. X., Liu, H., Fan, L., Li, L., Su, L. Q., & Wang, X. L. (2022). Effect of the phthalates exposure on sex steroid hormones in the US population. *Ecotoxicology and Environmental Safety*, *231*, 113203.

Zucker, K. J., Mitchell, J. N., Bradley, S. J., Tkachuk, J., Cantor, J. M., & Allin, S. M. (2006). The Recalled Childhood Gender Identity/Gender Role Questionnaire: psychometric properties. *Sex Roles: A Journal of Research, 54*(7-8), 469–483.

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