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**FEMALE AUTISM PHENOTYPES:  
SEX/GENDER DIFFERENCES IN  
FUNCTIONING, CAMOUFLAGING, AND  
EATING PROBLEMS**

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# Female Autism Phenotypes: Sex/Gender Differences in Functioning, Camouflaging, and Eating Problems

## Thesis for Doctoral Degree (Ph.D.)

By

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*Till Rebecca och Ester*



# Popular science summary of the thesis

Autism is a neurodevelopmental condition characterized by challenges in social communication and interaction alongside restricted and repetitive behaviors and interests and atypical sensory processing, causing disability in important areas of life. Historically, autism has been understood as a “male condition” rarely observed in girls and women. However, more recent research suggests that autistic females run the risk of having their autism going undetected and not receiving a diagnosis until later in life. In addition to the risk of late diagnosis leading to delayed support, the risk of other harmful outcomes including suicide and mental health problems such as anxiety and depression may be heightened among females on the autism spectrum. Still, further research is needed to advance our understanding of how autism may be differently expressed in females and males including risk factors for negative outcomes among autistic females.

Eating problems (e.g., issues driven by sensitivity to smell and texture of food) and camouflaging (strategies to mask or compensate for autistic traits) are two areas that could constitute risk factors for negative outcomes among autistic people and are both suggested to be especially common in autistic females. To date, research has established that eating problems are frequent among autistic children, but studies investigating if the link between autism and eating problems differs between females and males are scarce. Also, use of camouflaging is correlated to lower quality of life, depression, and even suicidality, suggesting that camouflaging is a potential risk factor that could be relevant to address in health care and by developing interventions. Still, no measure of camouflaging has been available in Sweden, hindering further research in this area. In addition, research has currently not established why camouflaging is correlated with quality of life and mental health problems. It may be that the correlations are due to camouflaging having a causal negative impact on these factors as is often hypothesized, but the correlations may also be due to influence from a third, confounding variable. The possibility of confounding must be ruled out before camouflaging can be pinpointed as a risk factor for negative outcomes.

The aim of this thesis was to contribute to knowledge about how autism may be differentially expressed in females compared to males, including differences in exposure to risk factors for harmful outcomes. These objectives were addressed by surveying the perception of sex/gender differences among professionals working with autistic people, and by collecting self-reports of eating problems and camouflaging from autistic and non-autistic people.

The results in **study I** showed that professionals working with autistic people perceived that autism was more difficult to recognize in females because the challenges in this group were seen as less apparent and due to use of camouflaging strategies further masking difficulties. In **study II**, the investigation of eating problems in a sample of

adolescent and adult twins showed that females with elevated levels of autistic traits were particularly at risk for overall eating problems and for experiencing difficulties eating with others, such as in school and at work.

In **study III**, a Swedish version of the Camouflaging Autistic Traits Questionnaire (CAT-Q) was developed and evaluated, which gave preliminary support for using the camouflaging measure in Swedish settings, but also indicated that further examination of the questionnaire is needed. While camouflaging behaviors were found also in the general population, use of such strategies appears to decrease during adulthood among non-autistic people but remain at high levels among autistic people. This difference could reflect that autistic individuals feel pressured to camouflage in order to be accepted in society, whereas non-autistic people use similar behaviors more voluntarily. Furthermore, the results in **study IV** showed that the association between camouflaging and lower quality of life remained also when comparing twins to each other within twin pairs, which shows that the link remains even when applying strict control of confounding factors.

In conclusion, the findings in this thesis point towards areas that may contribute to negative outcomes for autistic females, including delayed diagnosis, decreased participation in social interaction, and decreased quality of life. Both eating problems and camouflaging were linked to autism in females, while it should be noted that also some autistic males displayed such issues extensively. The results further strengthen the previous claim that camouflaging has a causal negative effect on quality of life. The findings in this thesis point towards the need for further research into both the consequences and management of eating problems and camouflaging, and that both should be addressed in health care settings to support everyday social functioning and quality of life for autistic females.



# Abstract

## Background

Autism is a neurodevelopmental condition defined by challenges in social interaction and communication and presence of repetitive patterns in behavior, interests, and activities alongside sensory processing alterations (RRBI). While a long-standing tenet of autism is a male predominance of the condition, recent research evidence indicate that autistic females are diagnosed late or even missed, limiting timely access to support, and potentially increasing the risk of mental health problems and reduced quality of life (QoL). Evidence also suggests elevated risks of other detrimental outcomes among autistic females, including suicide and internalizing mental health problems, although findings regarding the latter have been inconsistent. Research further characterizing female autism phenotypes and challenges in this group is warranted.

## Aims

The overarching aim of this thesis was to contribute to deeper insights into female autism phenotypes and associated risk factors for detrimental outcomes, via qualitative exploration of the perception of sex/gender differences in autism among professionals working with autistic people, and via quantitative investigations of eating problems and camouflaging.

## Methods

An expert survey exploring the perceptions of functional sex/gender differences in autism in a multi-disciplinary and international sample of professionals was conducted including descriptions of sex/gender-related characteristics pertaining to the individual as well as the environment. In addition, the link between dimensional and categorical operationalizations of autism and eating problems was evaluated including sex/gender moderation of the association, in a thoroughly phenotyped twin sample. Furthermore, a psychometric evaluation of a Swedish version of the Camouflaging Autistic Traits Questionnaire (CAT-Q) was performed, allowing further investigation of the construct. Finally, the hypothesized negative causal effect of camouflaging on QoL was tested using a co-twin control design and within-pair analyses controlling for familial confounding.

## Results

Professionals perceived autism manifestations in females as more difficult to recognize due to overall less salient challenges and increased use of behavioral strategies that camouflage autistic characteristics in this group. The investigation of eating problems indicate that problems such as selective eating and sensory issues in mealtimes are present among autistic adolescents and adults, but the association between autistic

traits and overall eating problems was especially pronounced in females. Also, eating with others in social situations arose as a specific difficulty experienced by autistic females. The psychometric evaluation of the Swedish version of the CAT-Q (CAT-Q/SE) yielded partial support for the reliability and validity of the scale and indicated that camouflaging behaviors may decline with increasing age in non-autistic people while remaining at elevated levels among autistic people. Camouflaging was significantly associated with reduced QoL, also when controlling for familial factors within dizygotic (DZ) and monozygotic (MZ) twin pairs, consistent with a causal effect.

## **Conclusions**

This thesis found support for sex/gender moderation in areas that are potentially involved in shaping important outcomes among autistic females, including delayed diagnosis, low social participation, and QoL. The findings support that eating problems and camouflaging are prominent in female autism phenotypes and strengthen previous claims of camouflaging strategies having a negative impact on QoL. Addressing eating problems and camouflaging in health care services may facilitate functioning and QoL and could be of relevance among autistic females.

# List of scientific papers

- I. **Lundin, K.,** Mahdi, S., Isaksson, J., & Bölte, S. (2021). Functional gender differences in autism: An international, multidisciplinary expert survey using the International Classification of Functioning, Disability, and Health model. *Autism, 25*(4), 1020–1035.
- II. **Lundin Remnélius, K.,** Neufeld, J., Isaksson, J., & Bölte, S. (2022). Eating Problems in Autistic Females and Males: A Co-twin Control Study. *Journal of autism and developmental disorders, 52*, 3153–3168.
- III. **Lundin Remnélius, K.,** & Bölte, S. (2023). Camouflaging in Autism: Age Effects and Cross-Cultural Validation of the Camouflaging Autistic Traits Questionnaire (CAT-Q). *Journal of Autism and Developmental Disorders, 1–16*.
- IV. **Lundin Remnélius, K.,** Neufeld, J., Isaksson, J., & Bölte, S. Does camouflaging cause reduced quality of life? A co-twin control study. [Manuscript in preparation].

## Scientific papers not included in the thesis

- I. Isaksson, J., Van't Westeinde, A., Cauvet, É., Kuja-Halkola, R., **Lundin, K.**, Neufeld, J., Willfors, C., & Bölte, S. (2019). Social cognition in autism and other neurodevelopmental disorders: a co-twin control study. *Journal of autism and developmental disorders*, 49, 2838–2848.
- II. Isaksson, J., Taylor, M. J., **Lundin, K.**, Neufeld, J., & Bölte, S. (2019). Familial confounding on the ability to read minds: A co-twin control study. *Autism*, 23(8), 1948–1956.
- III. Neufeld, J., Hagström, A., Van't Westeinde, A., **Lundin, K.**, Cauvet, É., Willfors, C., Isaksson, J., Lichtenstein, P., & Bölte, S. (2020). Global and local visual processing in autism—a co-twin-control study. *Journal of Child Psychology and Psychiatry*, 61(4), 470–479.
- IV. Myers, L., Anderlid, B. M., Nordgren, A., **Lundin, K.**, Kuja-Halkola, R., Tammimies, K., & Bölte, S. (2020). Clinical versus automated assessments of morphological variants in twins with and without neurodevelopmental disorders. *American Journal of Medical Genetics Part A*, 182(5), 1177–1189.
- V. Myers, L., Ho, M. L., Cauvet, E., **Lundin, K.**, Carlsson, T., Kuja-Halkola, R., Tammimies, K., & Bölte, S. (2020). Actionable and incidental neuroradiological findings in twins with neurodevelopmental disorders. *Scientific Reports*, 10(1), 22417.
- VI. Neufeld, J., Hederos Eriksson, L., Hammarsten, R., **Lundin Remnélius, K.**, Tillmann, J., Isaksson, J., & Bölte, S. (2021). The impact of atypical sensory processing on adaptive functioning within and beyond autism: The role of familial factors. *Autism*, 25(8), 2341–2355.
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- VIII. Neufeld, J., Taylor, M. J., **Lundin Remnélius, K.**, Isaksson, J., Lichtenstein, P., & Bölte, S. (2021). A co-twin-control study of altered sensory processing in autism. *Autism*, 25(5), 1422–1432.
- IX. Myers, L., Pan, P. Y., **Lundin Remnélius, K.**, Neufeld, J., Marschik, P. B., Jonsson, U., & Bölte, S. (2021). Behavioral and biological divergence in monozygotic twin pairs discordant for autism phenotypes: A systematic review. *JCPP Advances*, 1(2), e12017.
- X. Isaksson, J., Ruchkin, V., Aho, N., **Lundin Remnélius, K.**, Marschik, P. B., & Bölte, S. (2022). Nonshared environmental factors in the aetiology of autism and other neurodevelopmental conditions: a monozygotic co-twin control study. *Molecular autism*, 13(1), 8.

- XI. Austin, C., Curtin, P., Arora, M., Reichenberg, A., Curtin, A., Iwai-Shimada, M., Wright, R. O., Wright, R. J., **Lundin Remnélius, K.**, Isaksson, J., Bölte, S., & Nakayama, S. F. (2022). Elemental dynamics in hair accurately predict future autism spectrum disorder diagnosis: an international multi-center study. *Journal of clinical medicine*, *11*(23), 7154.
- XII. Carlsson, T., Rosenqvist, M., Butwicka, A., Larsson, H., Lundström, S., Pan, P. Y., **Lundin Remnélius, K.**, Taylor, M. J., & Bölte, S. (2022). Association of cumulative early medical factors with autism and autistic symptoms in a population-based twin sample. *Translational Psychiatry*, *12*(1), 73.
- XIII. Dumitriu, D., Baldwin, E., Coenen, R. J., Hammond, L. A., Peterka, D. S., Heilbrun, L., Frye, R. E., Palmer, R., Norrman, H. N., Fridell, A., **Lundin Remnélius, K.**, Isaksson, J., Austin, C., Curtin, P., Bölte, S., & Arora, M. (2023). Deciduous tooth biomarkers reveal atypical fetal inflammatory regulation in autism spectrum disorder. *Iscience*, *26*(3).
- XIV. Curtin, P., Neufeld, J., Curtin, A., Austin, C., Isaksson, J., **Lundin Remnélius, K.**, Norrman, H. N., Arora, M., & Bölte, S. (2023). Associations between Elemental Metabolic Dynamics and Default Mode Network Functional Connectivity Are Altered in Autism. *Journal of clinical medicine*, *12*(3), 1022.



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## List of abbreviations

ABAS-II	Adaptive Behavior Assessment System – Second Edition
ADHD	Attention-Deficit Hyperactivity Disorder
ADI-R	Autism Diagnostic Interview Revised
ADOS	Autism Diagnostic Observation Schedule
AN	Anorexia Nervosa
ANOVA	Analyses of Variance
AQ-10	Autism Spectrum Quotient-10
ARFID	Avoidant/Restrictive Food Intake Disorder
AUC	Area Under the Curve
CAT-Q	Camouflaging Autistic Traits Questionnaire
CAT-Q/SE	Camouflaging Autistic Traits Questionnaire, Swedish version
CATSS	Child and Adolescent Twin Study in Sweden
CFA	Confirmatory Factor Analysis
CI	Confidence Interval
CNV	Copy Number Variant
DSM-5	Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition
DZ	Dizygotic
DWLS	Diagonally Weighted Least Squares
EFA	Exploratory Factor Analysis
GAD-7	Generalized Anxiety Disorder 7-item scale
GAI	General Ability Index
GEE	Generalized Estimating Equations
HIC	High-Income Country
ICD-11	International Classification of Diseases
ICF	International Classification of Functioning, Disability, and Health
IQ	Intelligence Quotient

K-SADS	Kiddie Schedule for Affective Disorders and Schizophrenia
LOESS	Locally Estimated Scatterplot Smoothing
MIC	Middle-Income Country
MZ	Monozygotic
NSE	Non-Shared Environmental
PDD-NOS	Pervasive Developmental Disorder not otherwise specified
PHQ-9	Patient Health Questionnaire 9-item scale
PSS-14	Perceived Stress Scale
QoL	Quality of life
RATSS	Roots of Autism and ADHD Twin Study in Sweden
RMSEA	Root Mean Square Error of Approximation
ROC	Receiver Operating Characteristic
RRBI	Restricted and Repetitive Patterns in Behavior, Interests and Activities
SCID	the Structured Clinical Interview for DSM-IV
SiC	Stress in Children
SRMR	Standardized Root Mean Square Residual
SRS-2	Social Responsiveness Scale, Second Edition
SWEAA	Swedish Eating Assessment for Autism spectrum disorders
WAIS-IV	Wechsler Adult Intelligence Scale – Fourth Edition
WHO	World Health Organization
WHODAS	WHO Disability Assessment Schedule
WISC-IV	Wechsler Intelligence Scale for Children – Fourth Edition

# 1 Introduction

## 1.1 Autism

Autism spectrum disorder (henceforth *autism*<sup>1</sup>) is a neurodevelopmental condition defined by persistent social interaction and communication challenges in combination with restricted and repetitive patterns in behavior, interests and activities alongside sensory processing alterations (RRBI) (American Psychiatric Association, 2013), with a worldwide estimated prevalence in the range of 0.7% to 2.64% (Lyll et al., 2017). The prevalence of autism has seen a substantial increase since the condition was first described (Asperger, 1944; Kanner, 1943; Ssucharewa, 1926, 1927), which appears to not reflect an increase in autistic traits in the population, but instead likely involve a number of contributing factors such as changes in diagnostic criteria and increased awareness and recognition (Lai et al., 2014; Lundström et al., 2015). The condition is associated with detrimental outcomes, including limitations in relationships as well as in educational and vocational areas (Howlin & Moss, 2012), co-occurring conditions, premature mortality (Hirvikoski et al., 2016; Lai et al., 2014), and reduced quality of life (Jonsson et al., 2017; van Heijst & Geurts, 2015). Often dubbed a heterogenous condition, autism encompasses the full span of intellectual abilities, verbal abilities ranging from minimally verbal or non-speaking to fluent speech, as well as variable developmental trajectories and outcomes (Lord et al., 2020). The current understanding of autism is that the condition represents the extreme right-end of a distribution of autistic traits across the general population (Constantino, 2011).

### 1.1.1 Diagnostic criteria and assessment

Whereas previous diagnostic manuals included multiple diagnoses on the autism spectrum, such as Autistic disorder, Atypical autism, Asperger's syndrome, and Pervasive Developmental Disorder not otherwise specified (PDD-NOS), these are consolidated into one diagnosis in the current editions of diagnostic manuals, the fifth edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-5) and the 11th revision of the International Classification of Diseases (ICD-11). According to the operationalization in the DSM-5, autism comprises behavioral manifestations in two cardinal domains. The A-criterion includes limitations in social reciprocity, non-verbal communication, as well as in developing, maintaining, and understanding relationships and adapting to social settings. The B-criterion describes RRBI including subdomains of repetitive movements, insistence on sameness, restricted interests, and sensory hyper-

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<sup>1</sup> Striving to avoid ableist language, the term autism will be used rather than autism spectrum disorder in this review. Similarly, the behavioral presentation of autism will be described as autism characteristics rather than symptoms, and identity-first language is used (i.e., autistic person). (Bottema-Beutel et al., 2021).

or hyporeactivity. An autism diagnosis requires that all of the diagnostic criteria under A and at least two of the B criteria are displayed (currently or historically), that autistic characteristics were present in the early developmental period, are currently causing clinically significant impairment across important areas of functioning, and that behavioral challenges are not better explained by other conditions.

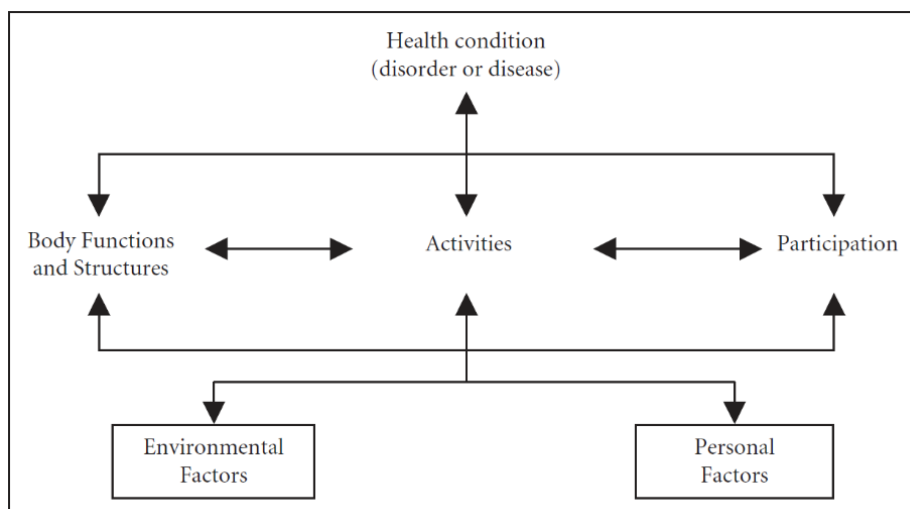
Autistic characteristics are usually persistent and early manifestations are observable after the first year of life, including reduced use of joint attention, increased repetitive use of objects and prolonged visual exploration (Zwaigenbaum et al., 2015). Additionally, regression, i.e., loss of previously acquired skills such as use of language or social interaction skills may occur in early development among as many as 32% of autistic individuals (Barger et al., 2013). However, early manifestations are heterogeneous, and autistic characteristics might not be fully manifest until later age when the social demands of the environment exceeds the individual's capacities (World Health Organization, 2018). Nevertheless, while many parents express concerns about their child in the early developmental period, most autistic people are diagnosed at a substantially later age (Bölte et al., 2020; Lord et al., 2020). As early identification of autism is associated with positive outcomes (Hosozawa et al., 2020; Mandy et al., 2022; Oredipe et al., 2022), further research aiming to facilitate reliable identification of autism and improve diagnostic assessment procedures is warranted.

A diagnosis of autism should be made based on clinician's overall evaluation of information from multiple sources, not relying on cut-offs from single measures (Bishop & Lord, 2023). Nevertheless, standardized instruments are helpful in collection of information relevant to autism, where the Autism Diagnostic Observation Schedule second edition (ADOS-2) and the Autism Diagnostic Interview Revised (ADI-R) are considered "gold-standard", and show balanced sensitivity and specificity when combined in assessments (Falkmer et al., 2013; Zander et al., 2015).

### **1.1.2 The International Classification of Functioning, Disability, and Health**

The diagnostic criteria outlined in the ICD-11 and DSM-5 are formulated from a biomedical perspective, acknowledging individual factors leading to impairment. While the diagnostic criteria are helpful in defining and operationalizing the condition, they do not yield a full and nuanced understanding of challenges and strengths among autistic people in their everyday lives and cannot capture the variety of functioning seen in the autism population. Given that factors pertaining to the environment such as access to support and autism-related stigma play important roles for functional outcomes in autism, any attempt to capture functioning among autistic individuals in a more comprehensive sense also need to take external influences into account (Bölte et al., 2021; Kirby et al., 2016). The International Classification of Functioning, Disability, and Health (ICF), is a framework for describing health-related functioning developed by the

World Health Organization (WHO, 2001). In the biopsychosocial perspective of the ICF, an individual's ability to engage in activities or tasks and participate in important areas of life are outcomes of both alterations in body structures and functions (including cognitive abilities) and contextual factors encompassing environmental and personal factors (e.g., sex/gender), see Figure 1. In the ICF framework, environmental factors encompass both physical, social, and attitudinal aspects of the environment, such as social norms, other people's attitudes, and access to support (WHO, 2007). Given that the ICF encompasses an extensive catalogue of functioning categories, short lists of the most relevant categories for specific health-conditions have been developed in order to facilitate the utility in clinical practice, and such Core Sets have been developed also for autism (Bölte, Mahdi, et al., 2019). Concerning the topic of focus in this thesis, the functional perspective of the ICF guides in acknowledging the interplay between individual strengths and limitations, sex/gender, and environmental factors in shaping the challenges faced by autistic females and males.



**Figure 1.** The ICF biopsychosocial model of functioning (WHO, 2007).

### 1.1.3 Etiology

Twin-studies have been an important source for insights into the etiological underpinnings of autism. Classical twin studies can disentangle the proportional contributions from genetics and environmental factors by comparing the degree of similarity between twins within monozygotic (MZ) pairs who are genetically identical and dizygotic (DZ) pairs who share on average 50% of their segregating genes. In twin designs, environmental factors are divided where shared environmental factors are non-genetic factors making twins within a pair similar, e.g. parental education, parenting styles, and amount of conflict in the family, and non-shared environmental (NSE) factors

on the other hand are environmental influences making twins dissimilar, such as medical events affecting one twin but not the other but also measurement error (Knopik et al., 2017). In classical twin studies, stronger correlations of autism and autistic traits between twins within MZ-pairs compared to DZ-pairs indicates a genetic influence, where heritability is defined the proportion of phenotypic variation that can be attributed to genetic differences among people (Knopik et al., 2017). A meta-analysis of twin-studies in autism reported substantial heritability estimates in the range 64% to 91% (Tick et al., 2016). Indeed, autism has been found to be one of the most heritable conditions (Wang et al., 2017). Findings from twin research also support that autism represents the extreme end of dimensional traits distributed continuously across the general population, as genetic factors associated with autism are also linked to lower levels of autistic traits (Lundström et al., 2012). The genetic underpinnings of autism are heterogeneous, where hundreds of gene variants have been linked to autism and involve contributions from rare mutations with substantial effects such as Copy Number Variants (CNVs), i.e., duplication or deletions of segments of the genome, as well as cumulative contributions of common variants with smaller effects (Vorstman et al., 2017).

While research evidence supports a substantial genetic influence in autism, the heritability estimates leave room also for environmental influences, where mainly NSE factors have been implicated (Bölte, Girdler, et al., 2019), and correlations between multiple tentative environmental influences and autism have been reported. However, environmental factors are often to a degree heritable themselves raising the possibility that the observed correlations reflect a genetic link between the variables (i.e., that common genetic factors influence both) rather than a causal impact of the environmental factor (D'Onofrio et al., 2013). Also, shared environmental influences could similarly confound the association, highlighting the need to establish a link between NSE factors and autism beyond such so called *familial confounding* from shared environment or genetics. Another twin design labelled *Co-twin control* adjust for familial confounding by comparing the association between exposure and outcome within differentially exposed twin pairs, comparing the twin exposed to a NSE factor to their unexposed co-twin, and thus holding all factors shared within twin pairs constant. Co-twin control studies suggest that NSE factors such as low birth weight and respiratory distress during birth may play a role in autism etiology (Carlsson et al., 2020), and point toward a potential cumulative effect of such environmental influences (Carlsson et al., 2022).

#### **1.1.4 Cognitive models of autism**

On the cognitive level, a range of theories have been purported to underpin the behavioral characteristics of autism. These can be divided into the so called social first theories and global processing theories (Lord et al., 2020). Social first theories purport that social cognition alterations form a core cognitive basis for autistic characteristics,

where limitations in theory of mind, i.e., “mindreading” capabilities – the ability to attribute mental states to others and oneself, is hypothesized as a domain-specific cognitive alteration leading to social interaction and communication challenges in autism (Baron-Cohen et al., 1985). A neighboring theoretical account suggests that social motivation, including the drive to orient attention towards social stimuli, being rewarded by social interaction, and striving to maintain social connections, is reduced in autism with cascading effects throughout development contributing to challenges in the social domain (Chevallier et al., 2012).

Global processing theories, such as the executive functioning account where cognitive limitations in goal-directed behaviors such as planning, impulse control, and cognitive flexibility are proposed to underpin the presence of RRBI but also altered social cognition in autism (Demetriou et al., 2018; Pellicano, 2007). Additionally, the local/global processing or weak central coherence account attempts to describe autism characteristics as arising from increased detail-orientation and reduced drive for global coherence (Frith & Happé, 1994). A neighboring account, the monotropism theory describes that autism is characterized by a narrow but intense scope of attention contributing to cognitive alterations, e.g., in cognitive flexibility and social motivation, as well as autism characteristics (Murray et al., 2005). While they have yielded support from research data, no single unifying cognitive theory of autism has been convincingly established. For example, cognitive atypicalities have not been replicated consistently in autistic samples, are not universally found among autistic people, and are often shared with other conditions such as attention-deficit hyperactivity disorder (ADHD) (Rajendran & Mitchell, 2007). In addition, social first theories have been questioned by accounts suggesting a *double empathy problem*, where mindreading difficulties are understood as being bidirectional in nature, arising in the interaction between autistic and non-autistic individuals where both struggle to understand the intentions and emotions of the other rather than constituting a deficit inherent in autistic individuals (Milton, 2012).

### **1.1.5 Co-occurring conditions and quality of life**

The majority of autistic people also display other neurodevelopmental, psychiatric, and somatic conditions. People on the autism spectrum are at higher risk for mental health problems than the general population, and around 70% of autistic people fulfill criteria for at least one co-occurring condition (Lai et al., 2014; Simonoff et al., 2008). More specifically, other neurodevelopmental conditions are frequent in autism, including intellectual disability and ADHD which are found in approximately 30% of autistic individuals respectively (Lai et al., 2019; Lyall et al., 2017). Other psychiatric conditions also commonly co-occur in autism, including anxiety disorders (reported among 20% of the autism population), depressive disorders (11%), and sleep-wake disorders (13%) (Lai et al., 2019). While prevalence estimates of co-occurring eating disorders are currently

scarce, findings support that eating disorders such as anorexia nervosa (AN) and avoidant/restrictive food intake disorder (ARFID) are more common in autism compared to the general population (Karjalainen et al., 2016; Nygren et al., 2021; Sedgewick et al., 2020). Autism is also associated with an increased risk of somatic conditions, including gastrointestinal problems (Muskens et al., 2017; Pan et al., 2019), and epilepsy, which is especially prevalent in autistic people with intellectual disability and among the most common causes of premature mortality in this subpopulation (Hirvikoski et al., 2016; Woolfenden et al., 2012).

Whereas autism in itself does not necessarily entail a negative impact on subjective wellbeing, co-occurring mental health problems contribute to reduced health-related quality of life (QoL) for autistic people (Adams et al., 2019; Bishop-Fitzpatrick et al., 2018; Helles et al., 2017). QoL is defined by the WHO as the individual's subjective appraisal of their position in life in relation to their goals, expectations, and standards (World Health Organization, 1998), and encompasses aspects of wellbeing that are related to or affected by a health condition (Coghill & Hodgkins, 2016). QoL is considered a multifaceted construct encompassing physical, psychological, social, and environmental domains (The Whoqol Group, 1998). Facilitating factors in the environment that have been related to higher QoL in autistic populations includes being diagnosed with autism early in life, having formal support needs met, and feeling that the available social support is sufficient for one's needs (Bishop-Fitzpatrick et al., 2018; Kamio et al., 2013; Renty & Roeyers, 2006).

## **1.2 Sex and gender**

One of the most well-established tenets of autism is that the condition shows a male predominance, which has sparked research interest in sex- and gender-related factors involved in etiology, phenotypic manifestations, assessment and diagnostic procedures, and outcomes. This research will be elaborated on later in this review, following a brief discussion on terminology and a (non-comprehensive) summary of sex- and gender-related findings in the general population.

The concept sex encompasses biological attributes in multiple layers, including chromosomal, gonadal, hormonal, internal reproductive, and genital components (Fausto-Sterling, 2012). A binary label of male or female sex is commonly assigned at birth based on a summary of these attributes that does not capture the entire range of the multileveled sex-related attributes which are not necessarily aligned (Bölte et al., 2023). *Gender*, on the other hand, is an umbrella term for socially constructed concepts, such as gender roles (gendered expectancies for responsibilities such as in households and workplaces), gender expression (the presentation of gender in everyday life), and sociocultural norms and expectations defining what is seen as appropriate behavior (Global Health 50/50, 2022). In addition, gender identity refers to an individual's sense



of their own gender which for some people is aligned with their sex assigned at birth and for some not (WHO, 2019a). This is of particular importance in the autism population, where experiences of gender diversity and not identifying with the sex assigned at birth are common (Dewinter et al., 2017; Strang et al., 2014).

While sex and gender are distinct constructs, they are interrelated and interact in shaping biology, behavior, and health outcomes (Bölte et al., 2023). Thus, in this literature review, the term sex/gender will be used to denote the interaction between sex- and gender-related attributes, where the effects rarely have been differentiated in the literature (Lai & Szatmari, 2020). Importantly, while neither sex nor gender are binary, the majority of research in sex/gender differences in autism include only males and females, which is reflected in this review.

### **1.2.1 Sex/gender differences in the general population**

Many findings of sex/gender moderation in behavioral or psychological constructs in the general population represent small differences with largely overlapping distributions among females and males (Fausto-Sterling, 2012). Examples of well-replicated differences include a female advantage in verbal fluency and theory of mind capacity, and an advantage in mental rotation tasks and increased physical aggression in males (Baron-Cohen et al., 2015; Fausto-Sterling, 2012; Hines, 2020; Hyde, 2014). As stated above, such differences may be influenced by both sex- and gender-related processes. According to the social cognitive theory of gender development (Bussey & Bandura, 1999), people learn about gendered norms and expectations from three sources: repeated observational learning from models (e.g., at home, in school, and in workplaces), the outcomes of their own behavior (other's evaluative reactions), and direct tuition (i.e., verbal descriptions of what is considered appropriate for boys and girls). Gendered behavior may thus be motivated by direct experiences and observational learning, and subsequently also by self-regulation informed by these experiences (Bussey & Bandura, 1999). Consistently, even when behaviors are associated with sex-related biological processes they may have interacting gender-related influences. For instance, sex/gender-typed choice of toys have been linked to prenatal testosterone exposure but also show social influences where play with gender-typical toys is rewarded by parents and peers (Hines, 2020). Similarly, aggression has been associated with prenatal testosterone but is also regulated via observational learning (Bölte et al., 2023; Bussey & Bandura, 1999). Importantly, research in this field has not only delineated a list of differences but suggests that sex and gender play important roles for health outcomes, including differences in the risk of health conditions, access to treatment, and treatment results (Mauvais-Jarvis et al., 2020).

## 1.3 Sex/gender in autism

### 1.3.1 Biological models of the male predominance in autism

As previously noted, autism is more often diagnosed in males than in females with an often cited male:female ratio of 4:1 (Fombonne, 2009; Zeidan et al., 2022). Aiming to explain the male preponderance in autism, theoretical accounts have suggested sex-related biological causes for the discrepancy. One of the most prominent models is the “*Extreme Male Brain*” theory which proposes that fetal testosterone exposure may be an important factor in autism etiology (Baron-Cohen, 2002). According to the theory, autism constitutes an extreme variant of certain cognitive characteristics found to be more pronounced in general population males compared to females. The theory stipulates that an average “male brain” is characterized by increased systemizing, i.e., a drive to construct and predict behaviors of systems, whereas a “female brain” on average show an increased drive to empathize, which includes attributing mental states to others (theory of mind) (Baron-Cohen, 2002). From a cognitive perspective, autism is then understood as an extreme variant of the “male brain”, which entails that only a small shift from the typical male cognitive profile is required for males to display an autistic phenotype, while the shift for females would be larger. The hypothesis that differential prenatal exposure to testosterone is linked to level of autistic traits, has yielded support in some studies (Auyeung et al., 2009), but not consistently (Bejerot et al., 2012; Kung et al., 2016).

A potentially complementary model is the “*Female Protective Effect*” (FPE) which purports that sex-related biological characteristics in females might decrease the likelihood of autism. Thus, the difference in autism prevalence is attributed to females requiring a larger etiological load (from genetic and environmental factors associated with autism) in order to display an autistic phenotype. This has been proposed to be due to sex-related protective effects, such as parent-of-origin imprinting of “protective” genes on the X-chromosome (Skuse, 2000). In support of the FPE hypothesis of an increased genetic load in autistic females, a larger proportion of autistic females display genetic mutations in form of de novo CNVs compared to autistic males, and also display larger CNVs (Levy et al., 2011). Support for the FPE model also come from familial aggregation studies. If the etiological threshold is higher in females, higher levels of autistic traits would be expected among the relatives of autistic females compared to relatives of autistic males. Research evidence supporting this hypothesis has been reported (Robinson et al., 2013), but is not found in all studies (Lai et al., 2015; Messinger et al., 2015). In summary, while the biological models have scientific merit and may contribute to the male overrepresentation in autism, current evidence is not sufficient for establishing that the male:female ratio is fully explained by biological causes.

### **1.3.2 Non-etiological explanations of the male:female ratio in autism**

In addition to the sex-related biological models, also non-etiological factors could influence the male:female ratio in autism, including that ascertainment of the condition is male-biased, making the interpretation of sex-related biological findings less clear as a proportion of autistic females might not have been included in the studies (Hull & Mandy, 2017). When investigated worldwide, the male:female ratio is not consistently observed across geographical regions but instead show substantial variation, suggesting that it might in part reflect differences in ascertainment and referral procedures (Elsabbagh et al., 2012; Zeidan et al., 2022). Consistently, while the 4:1 ratio is replicated in samples including people who already have a clinical diagnosis of autism, research studies utilizing active case-finding such as screening the general population report a less pronounced male dominance (closer to 3:1), implying that autistic females might be missed or identified later by clinical services (Loomes et al., 2017). Furthermore, a less pronounced ratio is found in adults compared to children, indicating that females are diagnosed later than males (Posserud et al., 2021), which is supported by a range of studies (Begeer et al., 2013; Giarelli et al., 2010; Shattuck et al., 2009). Consistently, in Stockholm, Sweden, the average age for a neurodevelopmental assessment yielding an autism diagnosis is nine years in boys and eleven in girls (Bölte et al., 2020).

### **1.3.3 Environmental barriers for diagnosis in females**

Delayed identification of autism in females could be influenced by gender-related environmental barriers in everyday settings such as in school as well as in gendered expectations and gender stereotypes on a societal level. School personnel can be important in initiating a first step towards neurodevelopmental assessment and autism diagnosis, functioning as “gatekeepers” to referral (Whitlock et al., 2020). However, teachers may not be as apt in identifying autism in girls who display a more female-typical autism phenotype (Whitlock et al., 2020), which will be described in detail later in this review. Correspondingly, teachers might miss challenges in girls even when parents rate that they have elevated autistic traits (Posserud et al., 2006), and are less concerned about social skills in autistic girls compared to autistic boys (Hiller et al., 2014). Also, the environment provided by non-autistic peers could make challenges in autistic girls less apparent to school personnel, where non-autistic boys often play organized games making it apparent when autistic boys are excluded, while autistic girls may give the impression of participating (e.g., by keeping in proximity of peers) in the more fluid social environment provided by non-autistic girls (Dean et al., 2017). Gender-related expectations and stereotypes around autism could also influence how autistic characteristics in females are viewed. For example, autism may be perceived as a “male condition”, making parents and clinicians less inclined to interpret behaviors in girls as manifestations of autism (Lockwood Estrin et al., 2021). Furthermore, social communication challenges in girls might be interpreted as gender-normative

characteristics such as just being “shy” rather than being linked to autism (Amr et al., 2011).

As indicated by the above, professionals working in educational and clinical settings have key roles in the everyday lives of autistic people including in referral and diagnostic processes, and thus hold important perspectives on aspects of sex/gender differences in autism that could contribute to delayed identification of autistic females as well as other important outcomes. Still, to date, research exploring how autism professionals perceive sex/gender differences in autism is scarce.

#### **1.3.4 Female autism phenotypes**

Environmental barriers likely interact with sex/gender moderation of the behavioral presentation of autism, where a *female autism phenotype*, i.e., a differing and less readily detected manifestation in females compared to males has been suggested to contribute to the risk of delayed identification of autistic females (Hull & Mandy, 2017). In addition to late diagnosis, the risks of other detrimental outcomes might differ for autistic females and males further highlighting the need for research into autism manifestations and outcomes that acknowledges sex/gender influences. The following sections summarize findings indicating that female autism manifestations commonly show subtle differences compared to male presentations, and subsequently, findings of sex/gender influences on detrimental outcomes in autism.

Differences in cognition and behavior between females and males on the autism spectrum may reflect typical differences also found in the general population or could alternatively arise from an interaction between sex/gender and autism, suggesting a sex/gender moderation of the impact of the condition (Hull, Mandy, et al., 2017). Regarding the cardinal domains of autism, females and males diagnosed with autism commonly show equivalent ADOS and ADI-R scores when compared on the social-communication domain, but females generally show fewer RRBI (Kaat et al., 2020; Mandy et al., 2012; Tillmann et al., 2018; Van Wijngaarden-Cremers et al., 2014). These findings appear to reflect those in the general population, where females also display lower RRBI scores compared to males (Hull, Mandy, et al., 2017; Messinger et al., 2015). Considering RRBI subdomains, autistic males show increased repetitive behaviors and restricted interests, whereas no difference is found for insistence on sameness (Uljarević et al., 2022). In contrast to other RRBI, autistic females may struggle more with sensory sensitivity (Lai et al., 2011; Moseley et al., 2018), which is included in the RRBI diagnostic criteria since the introduction of the DSM-5 (American Psychiatric Association, 2013). Still, this finding has not been consistently replicated (Beggiato et al., 2017; Hiller et al., 2014), and warrants further investigation.

While sex/gender differences are often not found in the broad social-communication domain, more finely granulated investigations indicate that related social characteristics

may differ between autistic females and males. Autistic girls show increased social attention to faces compared to autistic boys, (Harrop et al., 2019), and are more similar to non-autistic peers regarding social motivation and friendship quality (Head et al., 2014; Sedgewick et al., 2016). While showing less difficulties in developing friendships, autistic girls struggle in maintaining and understanding friendships, for example in recognizing conflict (Hiller et al., 2014; Sedgewick et al., 2016). Also, autistic girls show less challenges regarding social reciprocity in conversations and activities, making them more similar to neurotypical boys and girls (Hiller et al., 2014; Wood-Downie et al., 2020). Interestingly, autistic females may be more prone to use non-verbal communication such as gestures when talking, but do not display the typical female advantage in interpreting other people's use of non-verbal communication found in the general population (Baron-Cohen et al., 2015; Hiller et al., 2014; Isaksson et al., 2019). Additionally, autistic girls are more inclined to describe mental states when telling a story (i.e., that a character "thinks" or "knows" something) than autistic boys, whereas no difference is found between neurotypical boys and girls (Boorse et al., 2019). In summary, even when fulfilling diagnostic criteria for autism, the presentation in females show subtle differences making them appear more similar to non-autistic peers likely reducing the likelihood of referral and diagnosis (Lai & Szatmari, 2020), highlighting that further delineation of female autism phenotypes are required for an increased understanding and recognition of autism in girls and women.

### **1.3.5 Sex/gender differences in co-occurring problems and premature mortality**

Accompanying the risk of delayed diagnostic timing, other unfavorable outcomes may differ between autistic females and their male counterparts. First, delayed diagnosis in itself constitutes a potential risk factor for later co-occurring issues, where qualitative reports from late-diagnosed autistic women link tardy diagnosis to negative consequences such as lacking support and tolerance from others (Bargiela et al., 2016). Correspondingly, recent quantitative studies report associations between later diagnosis and increased mental health problems (Hosozawa et al., 2020; Rødgaard et al., 2021), where longitudinal data support that internalizing and externalizing problems increase at a higher rate among children diagnosed later when compared to children diagnosed by age seven, which could speculatively be mitigated with timely diagnosis and support (Mandy et al., 2022). Also, people who learn about being autistic in younger age generally report higher QoL in adulthood compared to their peers who learned that they were autistic later in life (Oredipe et al., 2022).

Regarding mental health problems, adolescent and adult females in the general population are at increased risk for internalizing psychiatric problems, such as depression and anxiety disorders (Rutter, Caspi, et al., 2003; Van Oort et al., 2009). In the autism population, research findings of sex/gender influences on co-occurring conditions have been mixed, where some studies find more internalizing problems in

autistic girls compared to autistic boys (Mandy et al., 2012), whereas others have not observed differences (Hudson et al., 2019; Simonoff et al., 2008). Two meta-analyses of depression in the autism population published in 2019 came to partially contrasting conclusions. Lai and colleagues (2019) reported higher rates of depression in studies including more females, whereas Hudson and colleagues (2019) did not find sex/gender moderation in the rate of depression, while noting that their findings could have been influenced by a substantial male predominance and young age in most samples. Notably, two recent, population-based studies in Scandinavian countries reported that autistic females were at increased risk for a multitude of psychiatric conditions, including anxiety and depressive disorders (Martini et al., 2022; Rødgaard et al., 2021). In these studies, several typical sex/gender differences were especially pronounced in autism, pointing towards a stronger link between autism and many co-occurring mental health problems in females.

The all-cause risk of premature mortality is elevated in both autistic males and females compared to the general population (Catalá-López et al., 2022). However, sex/gender differences are found when investigating specific causes. For example, autistic males may be at relatively higher mortality risk from diseases of the nervous and circulatory systems (Hirvikoski et al., 2016). On the other hand, autism is also associated to increased risk of suicide, which is pronounced in autistic women (Kirby et al., 2019), and at the highest levels among females with co-occurring ADHD but without intellectual disability where approximately 20% have been reported to have conducted at least one suicide attempt (Hirvikoski et al., 2016, 2019). The risk of suicide thus shows a reversed sex/gender difference among autistic people compared to the general population where longstanding data show that suicide is more common among males (WHO, 2019b). The association between autism and suicidal behaviors is only partially mediated by psychiatric conditions suggesting that other risk and protective factors needs to be investigated to understand and prevent the elevated risk of suicidal behaviors in autistic females (Hirvikoski et al., 2019).

### **1.3.6 Eating problems**

In addition to the co-occurring issues and outcomes summarized above, autism is associated with issues around eating which may be moderated by sex/gender (Ledford & Gast, 2006; Spek et al., 2020). In autism, eating problems often revolve around selective eating (i.e., consuming only a limited variety of foods, sometimes based on sensory aspects such as color or texture), food refusal, inflexibility in mealtimes (where the environment is required to be set up a certain way in order for the individual to eat), and such problems are more common in autistic children than neurotypical children (Råstam, 2008; Sharp et al., 2013).

Autism is also associated with an increased risk of co-occurring clinical eating disorders. Researchers have suggested a link between autism and AN, an eating disorder defined by restrictive eating leading to significantly low weight combined with fear of gaining weight and a distorted appraisal of weight or body shape, and data suggest that AN is more common in the autistic population than in the general population (Karjalainen et al., 2016; Sedgewick et al., 2020). In addition, the eating disorder termed ARFID is also more common in autism. This eating disorder is operationalized as avoidance or limitation of food intake, due to lack of interest in food, avoidance of food based on sensory characteristics, and concerns about aversive consequences of eating resulting in significant weight loss, malnutrition, dependence on supplementary feeding, or interference with psychosocial functioning (American Psychiatric Association, 2013). ARFID has been found to co-occur with autism (Bourne et al., 2020), and a small-scale Swedish study found that 28% of included autistic preschool children fulfilled criteria for ARFID where most children in this group had been prescribed nutritional supplements and/or displayed psychosocial dysfunction due to food intake problems (Nygren et al., 2021). Even for autistic people who have not been diagnosed with an eating disorder, problems related to eating could have negative consequences. For example, eating problems among autistic children is linked to increased levels of parental stress as well as internalizing and externalizing problems (Marquenie et al., 2011; Postorino et al., 2015). Selective eating of high-calorie foods might also contribute to the elevated rate of obesity in autism (Flygare Wallén et al., 2018; Kinnaird et al., 2019; Zheng et al., 2017). Furthermore, the link between autistic traits and eating disorder behaviors may be partly mediated by selective eating, suggesting that selective eating may play a role in the development of eating disorders in autistic people (Carter Leno et al., 2022).

Issues around eating and mealtimes have mainly been studied in autistic children whereas research in older age groups is limited, but suggests that eating problems remain also in adulthood (Karlsson et al., 2013; Spek et al., 2020). However, there is a surprising dearth of studies investigating sex/gender influences on eating problems in autism, especially since as many as 48 to 89% of autistic children display such issues (Ledford & Gast, 2006), and well-established knowledge state that females in the general population are at greater risk for disordered eating behaviors and multiple eating disorders, including AN (Bulik et al., 2007; Qian et al., 2013; Solmi et al., 2021). Two recent exceptions to this shortage point towards sex/gender influences, where Van't Hof and colleagues (2020) found that autistic traits were associated with emotional undereating (i.e., reacting to emotional arousal by eating less) only in girls. In addition, Spek and team (2020) reported that autistic women endorsed problems with sensory sensitivity in mealtimes and of accepting only particular brands of food to higher extent than autistic men, who struggled more with fine-motor problems around eating. Whereas both autistic men and women reported more eating problems than their neurotypical peers, autistic women reported having problems in a wider range of areas associated with food

and eating than autistic men. While preliminary evidence suggest that elevated eating problems may continue into adult age and be linked to female autism manifestations, previous research has not accounted for confounding from other neurodevelopmental and psychiatric conditions commonly co-occurring in autism, that are known to also be associated with eating problems (Fursland & Watson, 2014; Gal et al., 2011; Kaisari et al., 2017; Meng & D'Arcy, 2015). Furthermore, whereas Spek and colleagues (2020) investigated sex/gender differences among autistic and non-autistic adults, group sizes were modest and the study did not assess the link between dimensional autistic traits and eating problems, limiting the statistical power.

Twin-studies have yielded insights regarding the underpinnings of eating problems in the general population, where food neophobia (refusing new foods) and food selectivity show substantial heritability (Cooke et al., 2007; Smith et al., 2017). Still, while the phenotypic link between autism and eating problems is firmly established, less is known regarding why the two are related. A Swedish twin study found that the relationship between autistic traits and disordered eating (operationalized as failure to gain weight and fear of gaining weight) in childhood was explained by a common shared environmental influence (Råstam et al., 2013). However, this study did not explore the relationship between autism and commonly found eating problems such as selective eating or sensory sensitivity in mealtimes, and the generalizability to such common eating issues in autism, and to older age groups, are not clear.

## **1.4 Camouflaging**

Recent research has purported social coping strategies in autism termed *camouflaging* as a tentative risk factor for mental health problems, diminished QoL, and delayed identification of autism (Bargiela et al., 2016; Hull, Petrides, et al., 2017). Such strategies have been suggested to be especially prominent among autistic females (Attwood, 2000; Lai & Baron-Cohen, 2015), and can be defined as conscious or unconscious behavioral strategies contributing to a “less autistic” presentation in social interaction. Camouflaging covers a range of behaviors, including mimicking other people’s social behavior, forcing oneself to make eye contact, and preparing and following “scripts” in conversations (Hull, Petrides, et al., 2017). Use of these strategies are likely common in autism, in a survey of autistic adults, 70% reported camouflaging either in certain situations or across different contexts (Cage & Troxell-Whitman, 2019). Still, the frequency of camouflaging in the autism population is not well understood and research in the subpopulation with co-occurring intellectual disability is lacking. Autistic people often report camouflaging their autistic traits out of necessity as they otherwise are met with lacking acceptance and even bullying by neurotypical peers (Cage & Troxell-Whitman, 2019; Hull, Petrides, et al., 2017). Correspondingly, camouflaging might be a common reaction to experiences of autism-related stigma (Perry et al., 2022). In addition, autistic adolescents and adults also report camouflaging in order to facilitate



socialization, develop and manage relationships, as well as to reach pragmatic goals such as obtaining a job (Hull, Petrides, et al., 2017; Tierney et al., 2016).

Compensation is a closely related theoretical concept, which in this review will be subsumed under the umbrella-term of camouflaging, similar to previous research (Hull et al., 2020). The compensation account suggests that autistic people may use domain-general cognitive abilities such as intellectual or executive functions to compensate for social cognition (theory of mind) alterations (Livingston & Happé, 2017). As domain-general resources are employed also for a range of non-social tasks, these compensative efforts are likely straining, which has been supported by qualitative accounts describing that camouflaging requires intense concentration, self-control, and close monitoring of both the situation and one's own behavior (Hull, Petrides, et al., 2017). Currently, the hypothesized link with general intelligence has not yielded consistent support from empirical data (Hull, Petrides, et al., 2021; Lai et al., 2017; Livingston et al., 2019). Yet, as noted above, research has mainly included participants with average to high IQ, restricting the likelihood of finding an association. Some support has been found for the link between camouflaging and executive functions (Hull, Petrides, et al., 2021; Lai et al., 2017), which could represent an alternative cognitive route underpinning camouflaging, for example in facilitating planning of social behaviors such as what to say beforehand, as well as monitoring and inhibiting unwanted behavior in social interaction (Livingston & Happé, 2017).

#### **1.4.1 Tentative consequences of camouflaging**

In addition to the first-hand reports of the effortful and exhausting nature of camouflaging, these behaviors have been suggested to delay identification of autistic females by for instance making it more difficult for teachers to see social communication challenges in autistic girls (Lockwood Estrin et al., 2021). Still, while higher levels of camouflaging are observed among autistic people diagnosed in adulthood compared to those diagnosed in childhood (McQuaid et al., 2022), this hypothesis has currently not been clearly confirmed by research. Qualitative reports from autistic people also link camouflaging strategies to negative outcomes such as doubts about one's own identity and about relationships formed when camouflaging (Bargiela et al., 2016; Hull, Petrides, et al., 2017). Correspondingly, cross-sectional data show that camouflaging is associated with perceived stress, symptoms of anxiety and depression, reduced subjective wellbeing, and even increased suicidal thoughts and actions (Cage & Troxell-Whitman, 2019; Cassidy et al., 2018; Hull et al., 2018; Hull, Levy, et al., 2021). However, while often interpreted as reflecting consequences of camouflaging, current evidence is based on correlational findings while causal relationships have not been established. Importantly, correlations reported in previous studies might reflect confounding, including from familial factors, and research in this field has not accounted for the variety of explanations other than a causal relationship which could have given

rise to the observed covariation between camouflaging and the proposed detrimental consequences. For example, findings of a negative association between camouflaging and wellbeing could reflect a common genetic influence on both constructs rather than a causal effect. In fact, genes are frequently found to have pleiotropic effects influencing several traits, and phenotypic associations between wellbeing and other traits including optimism and risk of psychopathology have been found to be largely explained by common genetic influences (Bartels et al., 2013; Mosing et al., 2009). Thus, such alternative non-causal explanations must be falsified before stronger claims of a detrimental causal impact of camouflaging on mental health and QoL outcomes can be made.

#### **1.4.2 Measures of camouflaging**

The psychometric operationalizations of camouflaging can be broadly categorized as self-report measures and so-called discrepancy measures. Pertaining to the former category, the Camouflaging Autistic Traits Questionnaire (CAT-Q) is a self-report measure tapping intention and experiences of such strategies in social interaction, yielding a total score as well as scores for subscales Compensation (strategies to actively compensate for social interaction and communication challenges), Masking (strategies to hide autistic traits), and Assimilation (attempts to blend in during social situations experienced as uncomfortable) (Hull et al., 2018). Discrepancy measures, on the other hand, utilizes well-used measures of autistic characteristics, operationalizing camouflaging as the discrepancy between “subjective/internal” self-reports of autistic traits and/or theory of mind capability and “external” presentation, often measured by clinician ratings on the ADOS (Lai et al., 2017; Livingston et al., 2019). A mismatch, e.g., high levels of self-reported autistic traits but low levels of clinician-rated autistic characteristics, is then used as a proxy measure of camouflaging. As discrepancy measures depend on clinician-ratings of observable behavior, they are likely to capture “successful” camouflaging, i.e., when autistic characteristics actually are masked, rather than the intention to camouflage (Hull et al., 2018). Convergent validity evaluations of camouflaging measures have found correlations ranging from small or medium (comparing self-reported CAT-Q and a discrepancy measure) to large (parent-reported CAT-Q compared with self-reported CAT-Q; parent-reported CAT-Q with discrepancy measure), indicating that further investigations of the construct of camouflaging and its operationalizations are needed (Hannon et al., 2022; Milner et al., 2022). To date, camouflaging research has been conducted mainly in a few English-speaking countries and reports from other parts of the world is largely lacking. Other autism assessment measures show cultural differences even between countries often understood as reasonably similar, such as the USA and European countries (Bölte et al., 2008), which could be even more pronounced regarding camouflaging due to its inherent emphasis on the interplay between the individual and the environment (Hannon et al., 2022). Thus,

cross-cultural validation of camouflaging measures is needed in order to provide camouflaging measures for future research and clinical purposes outside of English-speaking countries.

### **1.4.3 Sex/gender differences in camouflaging**

Research data has provided support for more extensive camouflaging among autistic girls and women compared to autistic males (Cassidy et al., 2018; Hull et al., 2019; Lai et al., 2017; Wood-Downie et al., 2020), yet not consistently (Cage & Troxell-Whitman, 2019; Livingston et al., 2020). Further investigations are also needed among autistic people who do not identify with the sex that was assigned at birth, where research evidence is currently limited by the small group sizes included in studies (Hull et al., 2019; McQuaid et al., 2022).

The suggested sex/gender difference in camouflaging among autistic people could have multiple causes. Executive functions, and more specifically, better signal-detection sensitivity, potentially reflecting the ability to carefully monitor the social environment, is associated to a discrepancy measure of camouflaging in autistic women only (Lai et al., 2017). Increased social motivation in autistic females compared to autistic males (Sedgewick et al., 2016), could also contribute to increased camouflaging, reflecting that such strategies are conducted partly to facilitate interaction with other people (Hull, Petrides, et al., 2017). Speculatively, given differences in gender-related expectations and social contexts, autistic girls might experience other consequences for displaying autistic characteristics which could contribute to an increased pressure to camouflage. For example, autistic girls who are deemed socially insensitive may face more severe social repercussions than autistic boys, as the contrast with gender-based expectations is larger (Kreiser & White, 2014).

### **1.4.4 Camouflaging among non-autistic people?**

While camouflaging was originally described among autistic people, the phenomenon is understood as a dimensional construct, arguably reflecting that autistic traits are distributed across the general population (Hull et al., 2018). Accordingly, also people who are not diagnosed with autism self-report camouflaging behaviors, and some to a high degree (Hull et al., 2018; Jedrzejewska & Dewey, 2022). This has sparked ongoing discussions regarding the extent that camouflaging is autism-specific, or if the construct rather should be understood as involving behaviors connected to broader psychopathology (Fombonne, 2020; Lai et al., 2021). However, despite suggestions of similar strategies in other conditions, including in ADHD (Sonuga-Barke et al., 2022), the extent of camouflaging in other clinical groups is currently not well-understood. One proposition is that camouflaging is a form of *impression management*, a term coined by sociologist Erving Goffman to describe universal human attempts to control other people's impressions to achieve a favorable presentation of oneself (Ai et al., 2022;

Goffman, 1959). Similar to camouflaging, impression management in the general population is motivated by the need for social belonging and acceptance, but also by the drive to attain goals, such as gaining approval, increasing self-esteem, and to develop a desired identity (Leary & Kowalski, 1990). Camouflaging in autism may constitute a form of impression management with some unique characteristics. For example, among autistic people these behaviors appear to be strongly motivated by necessity, e.g., to avoid being targeted or bullied, whereas similar behaviors among neurotypical people might be used more voluntarily in order to increase social favorability (Ai et al., 2022), and to emphasize certain aspects of one's identity rather than attempting to mask one's "true self" (Pearson & Rose, 2021). In addition, people on the autism spectrum might struggle more in decoding social norms and conventions in neurotypical contexts, making their impression management less intuitive and more dependent on inflexible heuristics, i.e., camouflaging strategies, rather than behavior being guided by more precisely attuned prior beliefs and inferences (Ai et al., 2022).

In conclusion, previous evidence points toward camouflaging as a tentative risk factor for detrimental outcomes in autism, and particularly in females where these strategies may be common. Still, research studying the link between camouflaging and proposed consequences has not accounted sufficiently for confounding. Further, research in camouflaging is currently restricted by our limited knowledge concerning the construct itself. To elucidate the construct of camouflaging potentially fruitful routes for research have been suggested (Lai et al., 2021). These include more nuanced comparisons of specific behaviors between autistic and non-autistic people which could give insights into aspects that are more autism-specific versus equivalent across groups. Similarly, exploration of developmental trajectories of camouflaging behaviors could advance the understanding of how use of such strategies differs between age groups and if trajectories differ between autistic and non-autistic people. Also, the link between other clinical groups and camouflaging could be studied to extend our knowledge regarding if camouflaging pertains mainly to autism or also to other neurodivergent and psychiatric populations.

## **1.5 Summary**

As stated in this literature review, sex/gender are involved in shaping outcomes in autism. Substantial research evidence show that females are diagnosed at a later age than males, where first-hand qualitative reports are corroborated by quantitative data suggesting that late diagnosis could have detrimental outcomes, impacting QoL and mental health problems. Regarding sex/gender differences in mental health outcomes, research has so far rendered mixed evidence with recent research implying even more pronounced sex/gender-influence in autism compared to the general population. Furthermore, the elevated risk of suicide in autism is particularly high in autistic females,

in contrast to the sex/gender difference found in the general population, pointing towards specific challenges faced by females on the autism spectrum.

The research findings summarized here provide a preliminary basis for further understanding of sex/gender in autism, which could be implemented in the development of sex/gender-informed autism assessment, treatments, and interventions aiming to prevent impacts on functioning, QoL, and mental health outcomes. Still, a number of limitations currently restrict the understanding of sex/gender influences in autism. Investigation in this field has predominantly focused on core social-communication and RRBI domains and related behaviors, while functional perspectives encompassing both individual challenges and strengths and environmental factors are lacking. Furthermore, not much is known concerning the generalization of findings in high-income countries (HICs) to other regions of the world.

Two areas that are associated with autism, eating problems and camouflaging, constitute tentative risk factors for differential detrimental outcomes in autistic females and males. Eating problems are common among autistic children, show associations with internalizing and externalizing problems, and may contribute to the development of eating disorders. Still, while sex/gender influences on disordered eating are well-established in the general population, the role of sex/gender in the relationship between autism and eating problems is currently understudied.

Camouflaging is suggested to be a facet of female autism manifestations, and has shown correlations with mental health problems, reduced wellbeing, and suicidality. However, research in this topic has been hampered by lack of cross-cultural validation of camouflaging measures and conceptual unclarity surrounding the construct, including whether camouflaging strategies are autism-specific or a form of typical impression management, and the developmental trajectories of camouflaging. Also, previous findings of correlations between camouflaging and reduced wellbeing have not accounted sufficiently for potential confounding from familial factors which therefore remain as alternative non-causal explanations that have not been falsified, hindering a causal interpretation.



## 2 Research aims

The overarching aim of this thesis was to gain deeper insight into female autism manifestations and the differing challenges faced by autistic females and males. More specifically, sex/gender-related influences in autism were studied through the perceptions of professionals working with autistic people, as well as via self-reported experiences of eating problems and camouflaging among autistic people, which all may play important roles in delaying identification of autistic females as well as for functioning, mental health, and QoL outcomes.

**Study I** utilized the functional perspective of the ICF to explore sex/gender differences in autism in a holistic sense encompassing both factors pertaining to the individual and environmental barriers and facilitators. The primary objective was to explore how an international sample of professionals working with autistic people perceived functional differences among autistic females and males using survey data and qualitative content analysis. The second aim was to examine whether areas of functioning showing sex/gender differences were covered in the comprehensive ICF Core Set for autism. These Core Sets were developed according to a standardized procedure and based on four preparatory studies including both autistic females and males. Nevertheless, their validity in capturing functioning areas that may be especially important for autistic females has not been explicitly investigated, which is of importance as the Core Sets for autism may play an important role in future assessment and development of measures of functioning and disability in autism (Bölte, 2023). The third objective was to explore differences among professionals working in HICs and middle-income countries (MICs) regarding the functional sex/gender differences reported, to allow evaluation of the generalizability of sex/gender findings which have previously mainly come from HICs.

**Study II** aimed to provide a deeper understanding of the link between autism and eating problems in adolescents and adults including sex/gender influences and exploration into why a phenotypic link is observed. The first aim was to investigate the association between autism (defined categorically as diagnosis and dimensionally as autistic traits) and eating problems, considering both total and specific issues around eating and mealtimes. The second aim was to evaluate potential sex/gender moderation of the association between autism and eating problems. The third aim was to explore the influence of shared environment and genetics on the association using a co-twin control design and within-pair analyses. To address the objectives, the study involved a twin sample from the Roots of Autism and ADHD Twin Study in Sweden (RATSS) enriched for neurodevelopmental conditions including autism.

The aim of **study III** was to develop and psychometrically evaluate a Swedish version of a self-report measure of camouflaging (the CAT-Q) following classical test theory and including item analyses, reliability assessments of internal consistency and test-retest

stability, as well as examination of diagnostic, concurrent, and factorial validity. This also incorporated separate assessments in a subsample of 10-to-15-year-olds to investigate the utility of the Swedish version of the CAT-Q (henceforth, CAT-Q/SE) in this age group. In addition, the study aimed to explore age-effects on camouflaging among autistic and non-autistic people.

In **study IV** the main aim was to follow up on previous correlational findings, including those reported in **study III**, and test the hypothesis that camouflaging has a causal impact on subjectively appraised QoL, utilizing the RATSS project and a co-twin control approach allowing adjustment for unmeasured familial factors which have not been accounted for in previous camouflaging research. Specifically, we aimed to first investigate the association between autism and camouflaging, as well as potential sex/gender moderation of the link. The second aim was to assess the relationship between camouflaging and QoL across the sample while adjusting for potential confounding from sex, age, autistic traits, and ADHD. Third, the main objective, was to assess the association within DZ and MZ twin pairs providing implicit control for familial confounding. We expected a negative association between camouflaging and QoL across individuals, and that the association would survive adjustment for familial factors within DZ and MZ pairs, consistent with a causal effect.



## 3 Materials and methods

### 3.1 Design

#### 3.1.1 Study I

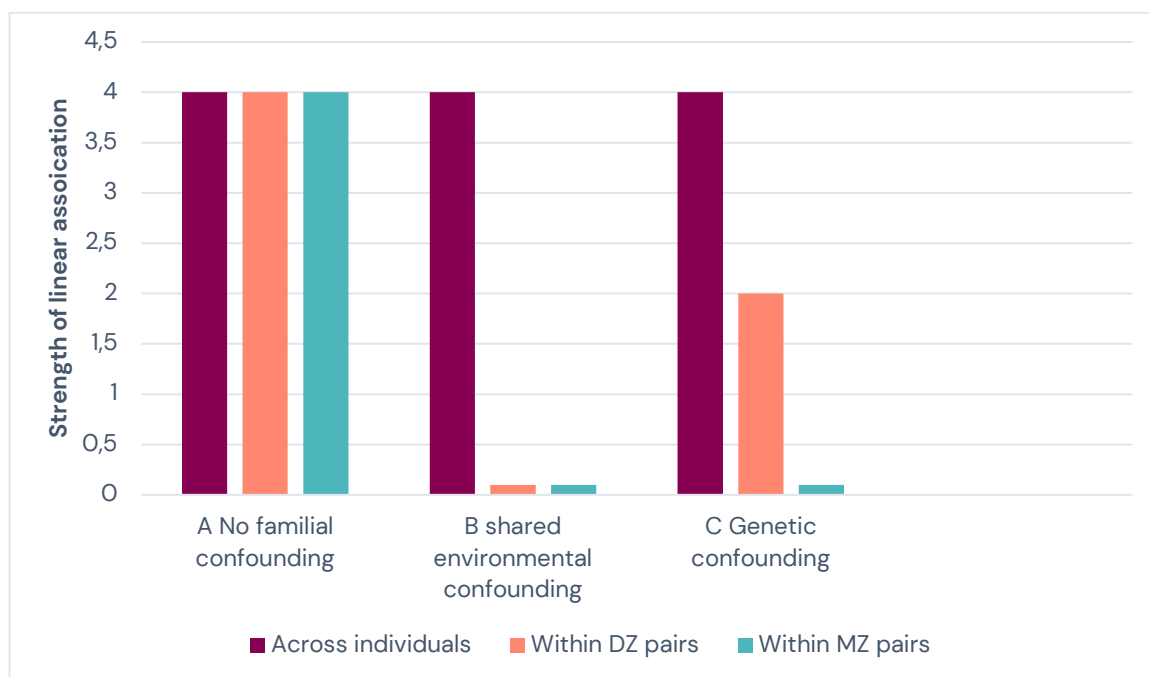
This was a qualitative study exploring responses to an open question on functional sex/gender differences in autism which was included in a larger expert survey into professionals' perspectives on functioning in autistic people conducted as part of the ICF Core Sets for autism project (Bölte, Mahdi, et al., 2019).

#### 3.1.2 Study II and IV

Both **study II and IV** utilized a co-twin control approach which is a powerful family-based design allowing investigation of the association between a putative risk factor and an outcome while accounting for familial factors, including shared environment and genetics (D'Onofrio et al., 2020). The co-twin control approach was applied in a two-step process: first, the association between exposure and outcome was assessed across individuals, similar to regression analyses commonly used in cross-sectional studies but with adjusted standard errors accounting for the clustering of the twin pairs. Second, the association was assessed within twin pairs testing if the association remains when adjusting for familial factors. For example, the within-pair analyses in **study IV** assessed if the twins displaying higher camouflaging also reported lower QoL compared to their co-twins. In the within-pair analyses, the twin-samples are stratified by zygosity, allowing comparison of the association in MZ pairs where shared environment and 100% of genetic factors are identical and thus controlled for, and in DZ pairs controlling for shared environment and on average 50% of segregating genes.

Figure 2 illustrates three hypothetical scenarios that could arise from the across individuals and within-pair analyses and how they are informative regarding the role of familial confounding. If the association between exposure and outcome across individuals remain within both DZ and MZ pairs the association cannot be due to familial confounding, which is consistent with a causal effect or an NSE influence on both variables (Figure 2, A). If the association is found across individuals but lost in both DZ and MZ pairs, findings are consistent with a shared environmental influence, which within-pair analyses in both DZ and MZ pairs fully control for, that likely largely drives the association (Figure 2, B). Finally, if an association is found across individuals and within DZ pairs but is lost within MZ pairs (Figure 2, C), findings are consistent with genetic confounding on the association. This interpretation assumes that the zygosity groups differ only regarding the level of genetics controlled for, as shared environmental factors are assumed to be equivalent in DZ and MZ pairs.

While the co-twin control design is powerful in testing putative causal or NSE influences on an outcome while controlling for familial factors, it can only give crude indications of the influences from these factors on an association. Thus, in contrast to classical twin designs using so called cross-twin cross-trait correlations, which assess the correlation between trait X in twin 1 and trait Y in twin 2, it will not yield estimations of the proportional contributions from NSE, shared environmental and genetic factors on a phenotypic association. Furthermore, the co-twin control approach cannot investigate prenatal or any other factors shared within twin pairs (D’Onofrio et al., 2013). Also, while the design can help ruling out confounding from familial factors, control for such factors is not sufficient for establishing a causal effect as confounding from other NSE and potential reversed causation are not accounted for by this design (McGue et al., 2010).



**Figure 2.** Hypothetical associations between exposure and outcome across individuals, within DZ pairs (controlling for shared environment and on average 50% of genetics), and within MZ pairs (controlling for shared environment and 100% of genetics). Scenario A is consistent with an association that is due to a causal effect or a NSE influence. Scenario B is consistent with a shared environmental influence on the association, and scenario C is consistent with an association that is due to genetic confounding. Adapted from Donahue et al (2013).

### 3.1.3 Study III

**Study III** was a cross-sectional psychometric evaluation of the CAT-Q/SE based on data from a web survey of autistic and general population samples. We aimed to include

participants representing a wide age range to permit investigation of the utility of the scale among 10- to 15-year-olds, and to explore age effects on camouflaging. In addition to the CAT-Q/SE, the web survey included demographic questions (e.g., gender identity, neurodevelopmental and psychiatric diagnoses, and age), and multiple other questionnaires measuring autistic traits, symptoms of depression and anxiety, perceived stress, QoL, and social functioning for planned analyses of concurrent validity.

## **3.2 Procedure**

### **3.2.1 Study I**

The expert survey was conducted between September 2013 and October 2014 as part of the process of developing ICF Core Sets for autism (de Schipper et al., 2016).

Recruitment was conducted via multiple routes, where contact information to eligible participants were collected from an international group of key opinion leaders in autism, as well as via academic, clinical, and community-based centers and university departments, and through an internet search. Snowball sampling was also used where experts were asked to recommend other eligible autism professionals.

The inclusion criteria required that the professionals practiced in one of the following professions: coach, counselor, nurse, occupational therapist, physician, physiotherapist, psychologist, psychotherapist, social worker, special educator, or speech-language pathologist. In addition, to be eligible for the study participants had to have at least five years of experience working with autistic people and be fluent in English.

### **3.2.2 Study II and IV**

The RATSS project (Bölte et al., 2014; Myers et al., 2021) was initiated in 2011 with ongoing recruitment until spring 2023. The majority of twins in RATSS were recruited from population-based twin-studies from the Swedish Twin Registry, where the Child and Adolescent Twin Study in Sweden (CATSS) (Anckarsäter et al., 2011) was the recruitment source for 46% of the full RATSS sample. Additional pairs were recruited via linking of Swedish Twin Registry data with other Swedish registries including the national patient registry of the National Board of Health and Welfare, clinical registries of the Child and Adolescent Psychiatry and the Habilitation and Health services. Participants were also recruited via adverts in publications from interest organizations (Society Attention, Autism Sweden) and through the RATSS webpage. Twins from population-based twin studies were prioritized if showing elevated scores on autism- or ADHD-scales, while participants with no previous indications of neurodevelopmental conditions were recruited as controls. It should however be noted that all participants' diagnostic status in RATSS was determined based on information collected during their participation, as detailed below.

In RATSS, exclusion criteria encompass profound intellectual disability, a serious psychiatric condition (e.g., schizophrenia), neurological conditions (e.g., intractable epilepsy), or presence of a well-defined genetic syndrome (such as fragile X). Included families participated in three days of assessments taking place at the Center of Neurodevelopmental Disorders at Karolinska Institutet (KIND) and at the Karolinska University Hospital. Data collection involved self- and parent-report questionnaires, psychiatric and neurodevelopmental assessments, cognitive testing, interviews, MRI scanning, and collection of biological samples including blood, saliva, hair, and stool. Participating families were assessed by three psychologists, where two conducted separate assessments with each of the twins, while the third psychologist led parent interviews. Best estimate clinical consensus DSM-5 diagnoses of neurodevelopmental and psychiatric conditions were based on the total information collected by the involved psychologists. Participating twins were categorized as MZ or DZ based on a panel of 48 single nucleotide polymorphisms (Hannelius et al., 2007), and for a minority of participants where genetic testing had not yet been conducted, by parent report on a 4-item questionnaire.

### **3.2.3 Study III**

The web survey was conducted from June 2020 to October 2020. Participants in Study III were included via two routes, where autistic participants were recruited from established contacts with interest organizations in Sweden (Autism Sweden, Society Attention, and Organized Aspergers). These participants completed a web survey hosted by Sunet Survey. In parallel, a geographically and demographically representative sample of the general population was recruited via a marketing and data collection company (PFM Research) from their preexisting panels. The general population participants answered the same questions on a web survey platform hosted by the marketing company. A subgroup in the general population sample self-reported having an autism diagnosis ( $n = 49$ ) and were included in the final autism sample.

## **3.3 Participants**

### **3.3.1 Study I**

Of the total 225 experts that responded to the survey, 131 provided a valid response to the question “In your experience with individuals with ASD, are there any aspects of their functioning and impairment that are specific to gender? If yes, please type your answer below”. Among the valid responses, 32 explicitly stated that they did not perceive gender differences in autism, and therefore the remaining 99 professionals were included in the qualitative analysis. These participants represented all six WHO regions (Africa, Eastern Mediterranean, Europe, the Americas, Southeast Asia, and Western Pacific), and 31 countries, where 71 professionals worked in HICs and 28 in MICs. Included professionals were predominantly female (77.8%), and most worked primarily in the

clinical field (45.5%), in education (15.2%), or in research (12.1%). The majority of the professionals worked predominantly with children and/or adolescents, whereas fewer worked primarily with adults.

### **3.3.2 Study II**

The final sample included 192 twins from the RATSS sample, comprising 63 MZ and 33 DZ pairs, aged 15 to 33 years (mean: 21.1 years), and 55% were female (based on sex assigned at birth). Twenty-eight participants were autistic (15 females and 13 males), 28 had ADHD, six mild intellectual disability, and 46 fulfilled diagnostic criteria for an internalizing psychiatric condition (e.g., social anxiety disorder, specific phobia, and major depression). These diagnostic categories overlapped, meaning that participants could be included in more than one group. In the sample, 16 twin pairs were categorically discordant for autism (7 MZ and 9 DZ pairs), and 59 pairs were defined as discordant for autistic traits (32 MZ and 27 DZ pairs), displaying intrapair differences greater than the reported standard error of measurement on the Social Responsiveness Scale-2 (SRS-2) total raw score (Constantino & Gruber, 2012).

In the total RATSS sample, 222 participants provided self-reported eating problems on the outcome measure, the Swedish Eating Assessment for Autism spectrum disorders (SWEAA). Of these, 30 participants were excluded resulting in the final 192 participants, due to one or both twins having completely missing data on any of the primary measures, or having moderate intellectual disability. Also, pairs that were of different sex or had pending zygosity were excluded, and so was one pair from a family where two twin pairs had participated in RATSS.

### **3.3.3 Study III**

The final sample encompassed 100 autistic participants and 539 general population (non-autistic) participants (N = 639). The autistic participants were aged between 10 and 68 years, and 55% identified as female. Regarding autism, 9% reported being diagnosed with autism before the age of 7, 28% in middle childhood (7 to 13 years), 12% in adolescence (13 to 18 years) and 50% in adulthood (> 18 years). Co-occurring conditions were common among autistic participants, where 50% reported a lifetime diagnosis of a co-occurring psychiatric condition, 41% ADHD, 5% intellectual disability, and 7% another neurodevelopmental condition. In the general population group, the age span was 10 to 83 years, and 61% identified as female. In this group, 21% reported a lifetime diagnosis of a psychiatric condition, 9% ADHD, 0.6% intellectual disability, and 6% another neurodevelopmental condition.

Of all individuals completing the web survey, 13 participants were excluded (10 from the general population and 3 autistic) due to straight lining throughout the questionnaires

(i.e., selecting the same response options for all items), yielding the final sample of 639 participants.

### 3.3.4 Study IV

The final sample in study IV included 140 individuals, representing 42 MZ and 28 DZ twin pairs participating in RATSS. The twins were aged from 10 to 41 years and 53% were female (sex assigned at birth). The sample included 22 autistic participants, 26 with ADHD, and 32 with at least one internalizing psychiatric condition (where participants can be included in more than one of these groups, e.g., have diagnoses of autism and ADHD). Among the participants, 28 MZ pairs and 21 DZ pairs showed at least an 8-point discordance for CAT-Q/SE total score, reflecting the standard error of measurement of the scale calculated from the results in **study III**.

Of the twin pairs in RATSS that had completed CAT-Q/SE and the QoL questionnaire used (EUROHIS-QOL), two pairs were excluded due to being different-sex pairs. In addition, we excluded five twin pairs where one or both twins fulfilled criteria for intellectual disability as we had experienced participants with low verbal abilities finding the CAT-Q/SE items challenging. Also, neither the English original version nor the Swedish version had been validated among people with intellectual disability. The exclusion of these pairs yielded the final sample of 140 individuals.

## 3.4 Measures

### 3.4.1 Measures used in RATSS

Twins included in **study II and IV** participated in an extensive battery of assessments as part of the RATSS protocol (Bölte et al., 2014). Diagnosis of autism was informed by gold-standard autism assessment instruments ADOS-Generic or 2nd edition (ADOS-G; ADOS-2) (Gotham et al., 2007; Lord et al., 2012) and ADI-R (Rutter, Le Couteur, et al., 2003). Diagnoses of other psychiatric and neurodevelopmental conditions, including ADHD, were supported by information from medical history and clinical interviews, including the Kiddie Schedule for Affective Disorders and Schizophrenia (K-SADS) (Kaufman et al., 1997), the Diagnostic Interview for ADHD in adults (Kooij, 2010), and the Structured Clinical Interview for DSM-IV (SCID, axis I). To measure IQ and support diagnosis of intellectual disability, the General Ability Index (GAI) from the fourth edition of the Wechsler Intelligence Scale for Children or the Wechsler Adult Intelligence Scale (WISC-IV; WAIS-IV) was used (Wechsler, 2003; Wechsler et al., 2008). In addition, adaptive behavior was assessed via parent report on the second edition of the Adaptive Behavior Assessment System (ABAS-II) (Harrison & Oakland, 2003).

In **study II**, eating problems were operationalized using self-report on the SWEAA, which was developed based on a literature review and clinical experience of eating problems

in autism (Karlsson et al., 2013; Råstam, 2008). The SWEAA yields a total score as well as subscale scores for eight subscales measuring specific eating issues such as sensory sensitivity in mealtimes (A. Perception), selective eating (D. Eating behavior), and behaviors related to clinical eating disorders (G. Other behavior associated with disturbed eating). The scale has shown acceptable reliability regarding internal consistency (all subscales  $> .73$ ), good test-retest stability (average subscale  $ICC = .860$ ), and diagnostic validity in differentiating between autistic and non-autistic people (Karlsson et al., 2013; Spek et al., 2020).

In **study II and IV**, autistic traits were measured via parent report on the child or adult versions of the Social Responsiveness Scale, second edition (SRS-2), which has shown excellent internal consistency ( $\alpha = .95$ ), indications of moderate to good test-retest reliability (.54 - .87), as well as acceptable-to-outstanding diagnostic validity ( $AUC = .79$  to .94) when comparing autistic and general population samples (Constantino & Gruber, 2012).

Camouflaging was measured via self-reports on the Swedish version of the CAT-Q, the CAT-Q/SE. The original CAT-Q encompass 25 items which were developed based on a qualitative study of camouflaging experiences among autistic adults (Hull, Petrides, et al., 2017). The items are answered on a seven-point Likert scale ranging from strongly disagree to strongly agree. Each item yields a score from one to seven, where five of the items are reverse worded and therefore reverse scored, resulting in a total score ranging between 25 to 175 with higher scores indicating increased camouflaging behaviors. The original UK version of the CAT-Q has shown a three-factor structure, and displays good internal consistency ( $\alpha = 0.94$ ) and acceptable test-retest reliability ( $r = .77$ ) (Hull et al., 2018). The original CAT-Q was translated from English to Swedish, where challenges arising in the process was reconciled via clarifications from the first author of the original validation study (Hull et al., 2018). After a first Swedish translation was completed, the scale was backtranslated by a professional translator and authorized after minor adjustments had been conducted. The CAT-Q/SE was thereafter piloted by experienced clinicians at a child and adolescent psychiatry center, and feedback was collected. Final adjustments were made where only the verbal anchors for the extreme values (1 and 7 on the Likert scale) were kept as the other verbal descriptions were reported being more confusing than helpful.

In **study III and IV**, QoL was measured using self-reports on the EUROHIS-QOL 8-item index which is an abbreviated scale derived from the WHO Quality of Life Brief Scale (WHOQOL-BREF) (Power et al., 2003). The eight items in EUROHIS-QOL cover subjective QoL in the psychological, social, physical, and environmental domain, and are answered using a five-point Likert scale. The EUROHIS-QOL has shown acceptable cross-cultural reliability and validity (da Rocha et al., 2012; Schmidt et al., 2006)

### 3.4.2 Other measures

The web survey in **study III** included a range of self-report questionnaires, in addition to the CAT-Q/SE and EUROHIS-QOL. Autistic traits were measured using self-report on the Autism Spectrum Quotient-10 (AQ-10), an abbreviated version of the full 50-item AQ. The AQ-10 is reported to show a unidimensional structure, sufficient reliability, and capacity to differentiate between autistic and neurotypical people (Allison et al., 2012; Lundin et al., 2019). Symptoms of depression were measured using the nine-item Patient Health Questionnaire (PHQ-9) (Kroenke et al., 2001), whereas anxiety symptoms were self-reported on the Generalized Anxiety Disorder seven-item scale (GAD-7) (Spitzer et al., 2006). Perceived stress was assessed using the Stress in Children (SiC) questionnaire for participants younger than 15 years (Osika et al., 2007), and on the Perceived Stress Scale (PSS-14) for participants 15 years or older (Cohen et al., 1983). Finally, social functioning was measured using self-reports on the Getting along subscale from the child and adult versions of the WHO Disability Assessment Schedule (WHODAS Child and WHODAS 2.0) which is based on the ICF framework (Canino et al., 2013; Scorza et al., 2013; Üstün et al., 2010).

## 3.5 Analysis

### 3.5.1 Study I

The written responses were analyzed using qualitative content analysis with an inductive approach, where categories were developed from the manifest level of the qualitative data (Bengtsson, 2016; Elo & Kyngäs, 2008). Following the process outlined by Bengtsson (2016), the qualitative material was first read through repeatedly, and subsequently sequences of text relevant to the primary research aim were labelled with codes. After controlling that no relevant data in the original responses were omitted, codes were clustered into categories, and categories showing similarity were grouped under main categories. Finally, categories were refined into a final structure. Addressing the second aim, the developed subcategories were linked to second-level ICF codes through a deductive process following linking rules from the ICF research branch (Cieza et al., 2002, 2005, 2019), allowing comparison with the comprehensive Core Set for autism. Finally, two-sided Fisher's exact test was used to compare the proportion of professionals from HICs and MICs referencing the main- and subcategories developed in the inductive content analysis.

### 3.5.2 Study II and IV

Statistical analyses in **study II and IV** followed similar procedures, where the associations between exposure and outcome were investigated using multiple linear regressions in the Generalized Estimating Equations (GEE) framework with cluster robust



standard errors accounting for the twins forming clusters rather than being independent data points (Zetterqvist & Sjölander, 2015).

The GEE is a fixed-effects model which allows regression analyses to be fitted across the sample treating participants as individual data points, similar to ordinary least squares regression models. The analytic procedure in **study II and IV** included first fitting GEE linear regressions across individuals assessing the association between exposure and outcome while accounting for twin-clustering. Second, conditional GEE models were applied to examine the within-pair association between exposure and outcome, while holding familial confounding factors constant. The conditional GEE can thus be used to evaluate if twins discordant for the exposure show differences in the outcome when all variables shared between twins within a pair are controlled for, including sex (in our studies which includes only same-sex DZ pairs), age, shared environment, and genetics. In the conditional within-pair analyses, the samples in **study II and IV** were split into MZ and DZ pairs as this gives an indication of how the association between exposure and outcome is impacted by controlling for ~50% of genes in DZ pairs and 100% in MZ pairs who are genetically identical.

These analyses require that only same-sex DZ pairs are included, as inclusion of DZ pairs discordant for sex assigned at birth would entail that comparisons within MZ pairs would control for sex while this would not be the case in DZ pairs, interfering with the assumption that degree of control for genetic confounding is the only factor separating the zygosity groups.

### **3.5.3 Study III**

The psychometric evaluation in this study was conducted in accordance with classical test theory. The CAT-Q/SE was analyzed on the item-level, and regarding reliability and validity on total scale level. The item analyses included standardized item difficulties evaluating the capacity to discriminate between individuals of separate items, corrected item-total correlations showing the alignment between separate items and the overall scale, and item validity evaluating the capacity of each item to differentiate between autistic and general population groups using non-parametric Wilcoxon rank sum tests.

*Reliability* was evaluated for the total scale and subscales regarding internal consistency calculated using Cronbach's alpha, as well as test-retest stability among 134 participants who completed the CAT-Q/SE a second time 4 to 10 weeks after submitting the web survey, using Pearson correlation coefficients and Intraclass Correlation Coefficients (ICC). Additionally, reliability was also determined separately in the subsample aged 10 to 14 years.

*Validity* was evaluated in terms of construct validity, including diagnostic validity, concurrent validity, and factorial validity. Diagnostic validity was calculated to evaluate if

the CAT-Q/SE differentiated between autistic and non-autistic groups and autistic females and males, as expected. Analyses of variance (ANOVA) was used to test the main effect of group (autism or general population) and the interaction effect autism by sex/gender, where interaction effects were followed up in the autism and general population groups separately using post hoc *t*-tests. Receiver operating characteristic (ROC) curves were also calculated to investigate how accurately the CAT-Q/SE total scores classified participants as autistic or non-autistic, in the full sample as well as separately for participants aged 10 to 14 years and 15 years or older. Concurrent validity was assessed by examining if the pattern of observed Pearson correlations between CAT-Q/SE scores and theoretically neighboring measures reflected the hypothesized pattern, see Table 1. Factorial validity was determined by conducting a Confirmatory factor analysis (CFA) evaluating the fit of the three-factor structure found for the original CAT-Q (Hull et al., 2018), using Diagonally Weighted Least Squares (DWLS) estimators and robust statistics (DiStefano & Morgan, 2014). If the CFA would indicate a lacking fit for the original factor structure, exploratory factor analysis (EFA) using Principal Axis Factoring with oblique (promax) rotation was conducted.

Finally, to explore the relationship between age and CAT-Q/SE scores in the autistic and non-autistic groups respectively, locally estimated scatterplot smoothing (LOESS) curves were fitted in each group. In addition, continuous norms for the autistic and general population were developed using polynomial regression models.

**Table 1.** Hypothesized pattern of associations between camouflaging and theoretically linked constructs for concurrent validity analysis (Lundin Remnélius & Bölte, 2023).

<b>Construct</b>	<b>Expected strength of the association</b>	<b>Theoretical relationship</b>
Autistic traits (AQ-10)	Moderate-to-large positive association	Autistic traits are linked to the core of the construct, camouflaging is suggested to encompass strategies to make existing elevated autistic traits less apparent (Hull et al., 2019).
Social functioning (Getting along subscale; WHODAS 2.0/WHODAS Child)	Moderate positive association (as higher WHODAS scores indicate more disability)	Lower social skills are believed to increase likelihood to use camouflaging (Hull et al., 2018), and such strategies might contribute to not getting the support needed, further decreasing social functioning (Livingston, Shah, et al., 2019)
Depressive symptoms (PHQ-9) Anxiety symptoms (GAD-7) Stress (PSS-14/SiC) Quality of Life (EUROHIS-QOL)	Small-to-moderate positive associations, except quality of life where a negative association is expected	Theorized as detrimental consequences of camouflaging, e.g., exhaustion, experiences of presenting a “false” self, anxiety concerning whether camouflaging was “successful”, and reduced access to support (Bargiela et al., 2016; Cassidy et al., 2020; Hull et al., 2017, 2021; Lai et al., 2017).

### 3.6 Ethical considerations

All studies were approved by the regional ethical review board in Stockholm or the Swedish Ethical Review Authority. For **study I**, ethical application and approval included the entire ICF Core Sets project (Dnr 2014/53-31/2). For **study II and IV** which were conducted within RATSS, ethical approval was obtained for the RATSS main application (Dnr 2016/1452-31) and a subsequent amendment allowing addition of the CAT-Q/SE and EUROHIS-QOL to the RATSS protocol (Dnr 2019-03279). For **study III** a separate main application was approved (Dnr 2019-06307).

In **study I**, eligible experts were contacted via an email entailing study information and those who agreed to participate were sent the survey. In **study II and IV**, study information was given first via telephone and subsequently in writing where families were given ample opportunities to ask potential questions before participation. Depending on the age of the twins, twins themselves and/or parents consented to the participation in RATSS. In **study III**, all participants gave informed consent before accessing the web survey, and parent consent was required for participants under the age of 15 years. Before accessing the survey and after the survey was completed, all participants were given contact information and were instructed to contact the research team with any questions.

Several aspects of this project warrant ethical considerations. First, the twins participating in the RATSS project represent a wide age range from children aged 8 years to adults aged 38 years, and a large proportion of the sample has neurodevelopmental or psychiatric diagnoses, including autism and ADHD. Study participation is extensive (Bölte et al., 2014), both regarding the time required and the activities which includes a range of assessments as detailed above, and can thus be demanding and energy-draining. Therefore, the team involved in the RATSS assessments strive for flexibility, attempting to adapt the procedure as much as possible to the specific participants and making room for breaks when needed, in order to minimize the strain on participating families. Another important aspect is ensuring that families feel that they can ask questions during their visit and that they can decline partaking in any assessments. To facilitate that families are comfortable throughout the RATSS visit, the same nurse or research assistant responsible for the first telephone contact follow the families between and often during assessments. Also, the psychologists responsible for the neurodevelopmental and psychiatric assessments are available for questions and for giving feedback to the families afterwards. In some instances, the assessments will indicate that participants with no former clinical diagnoses have unmet support needs and fulfill diagnostic criteria for neurodevelopmental or psychiatric conditions. In such cases the involved psychologists guide the families to appropriate clinical care, and if consent is given by the family, can also communicate information and relay test results to the responsible clinicians.

The topic of sex/gender differences in autism also relates to external ethical issues regarding how findings are communicated to the broad public. Sex/gender findings are generally on a group-level, where distributions in females and males are overlapping. Because of this, care must be taken to avoid creating new gender stereotypes by labelling traits more commonly displayed in one sex/gender as specifically “male” or “female”. Such labelling could mask important issues when displayed in other sex/genders (e.g., camouflaging among autistic males) and thus reflect previous problematic notions such as autism being “a male condition” which may have masked autism in females. Therefore, while research in this field is warranted to identify challenges and risks of harmful health outcomes for autistic females and their male counterparts, the overlap between and heterogeneity within sex and genders must be highlighted.



## 4 Results

### 4.1 Study I

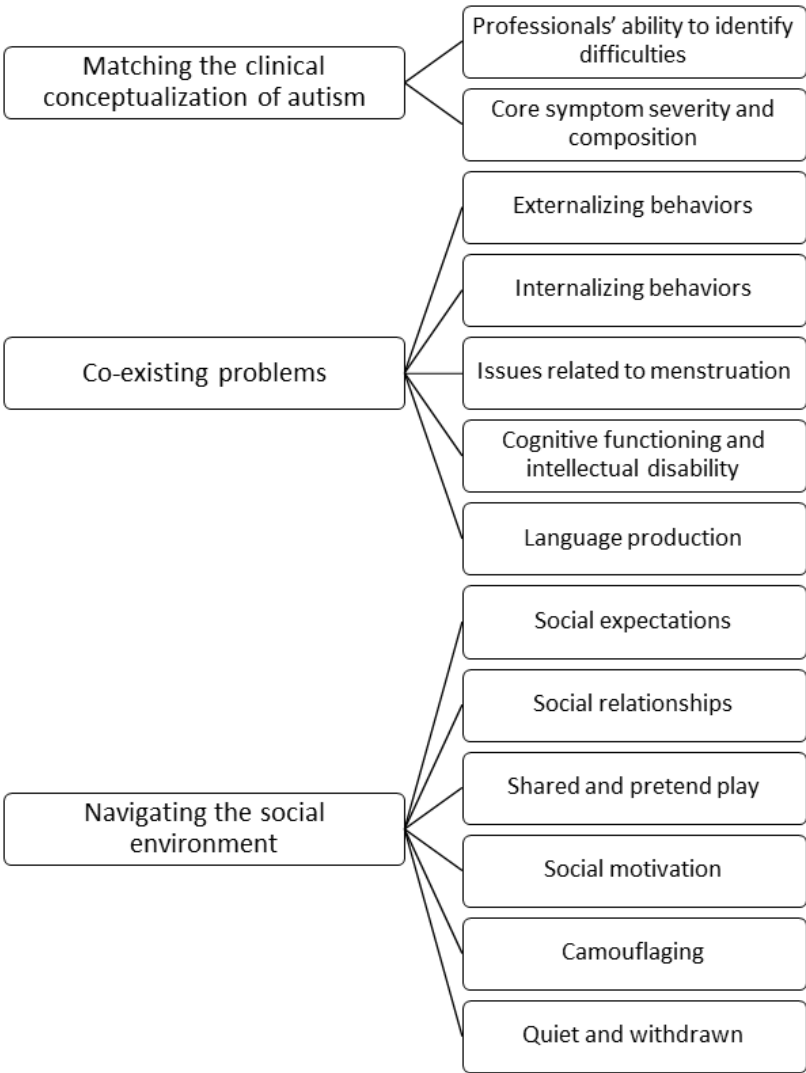
Three main categories and 13 subcategories were developed in the content analysis, capturing the professionals' perception of functional sex/gender differences in autism (see Figure 3). The first main category, *Matching the clinical conceptualization of autism* encompassed how professionals perceived that autism characteristics in females were more difficult to detect and less congruent with the criteria in diagnostic manuals. This main category also included references to differences in the manifestation and severity of core autism characteristics, e.g., that the focus of restrictive interests in autism often reflects typical sex/gender differences, where topics such as dolls and fashion were attributed to autistic girls, and that the focus of interests may be switched more often among autistic females.

The second main category captured references to *Co-existing problems*. Here, several professionals attributed autism in males with externalizing problems such as aggression and hyperactivity, which was perceived as contributing to social isolation in this group, whereas co-occurring internalizing problems were linked to female autism manifestations.

The third main category, *Navigating the social environment*, assembled descriptions of sex/gender differences in how autistic people manage their social surroundings, including both factors related to the individual and the environment. Professionals perceived that autistic females and males struggled with fulfilling gender-related norms in differing ways, where autistic females were described as facing higher expectations regarding social competence. Autistic males were perceived as being at greater risk for being bullied and rarely having friends supporting them. In contrast, autistic females were characterized as showing greater interest in social interaction and drive to develop relationships, which was linked to a risk of victimization and sexual abuse. Also, a mismatch arising from such social motivation and the social-communication challenges characterizing autism was reported to put autistic females at risk for experiences of "social failures" and distress. Furthermore, professionals perceived that environmental demands on social competence along with internal social motivation among autistic females underpinned the use of camouflaging strategies in this group, including copying behavior from peers and fictional characters and practicing facial expressions in the mirror. Such strategies were reported to give a surface-level impression of higher social competence, leading to the environment not adapting social expectations to the actual ability of the autistic girl or woman.

Regarding the second aim of the study, 31 of the 32 ICF categories that were linked to the subcategories were included in the comprehensive Core Set for autism. Further,

addressing the third aim, comparisons between professionals from HICs and MICs mainly revealed that similar proportions in both groups referenced most categories. However, the main category Navigating the social environment was referenced by a significantly larger proportion of participants from HICs than participants from MICs (52.1% vs 14.3%), which was also found for the subcategory Camouflaging (23.9% vs 0%). When comparing demographic characteristics between participants included in the content analysis ( $n = 99$ ), and participants responding that they did not perceive differences ( $n = 32$ ) or who did not provide a response ( $n = 91$ ), no significant group differences were found, e.g., regarding proportion of experts working in HICs, proportion of females, or years of experience.



**Figure 3.** Main categories and subcategories. (Lundin et al., 2021)

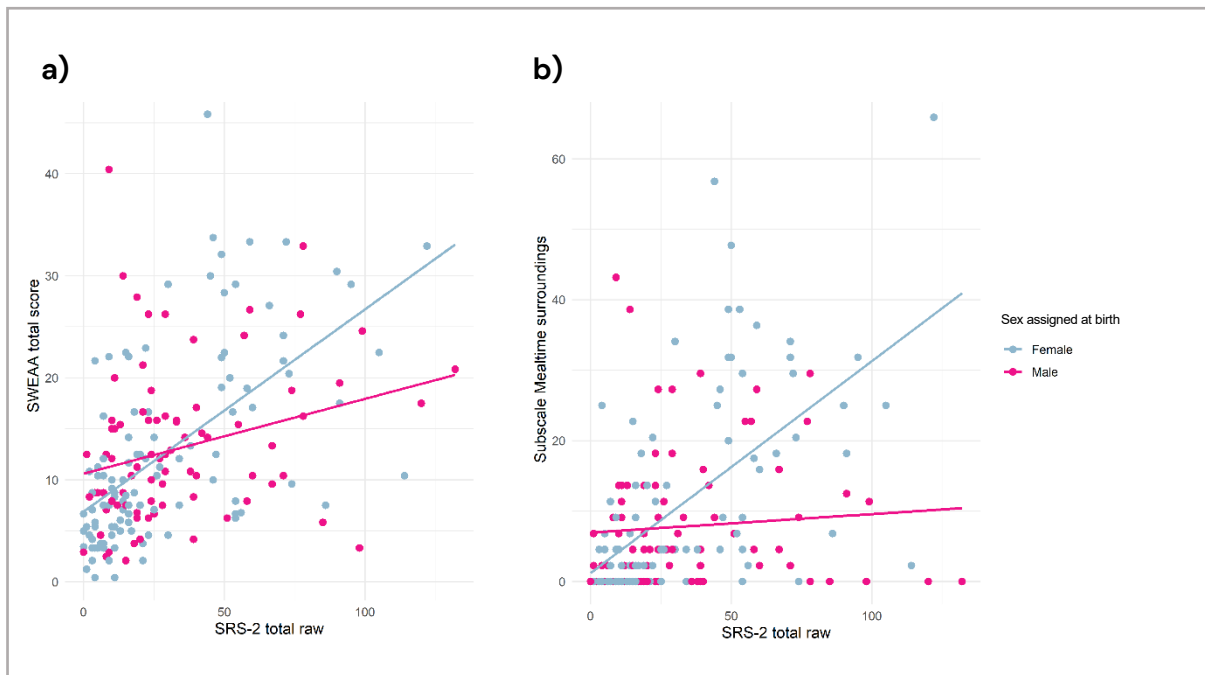


## 4.2 Study II

The results supported a link between autism and eating problems in adolescents and adults, as well as sex/gender moderation on the link. As predicted, both autistic traits and autism diagnosis were associated with total eating problems, also when adjusting for covariates sex, age, ADHD, internalizing conditions, and IQ (autistic traits:  $b = 0.17$  [95% *CI*, 0.11 to 0.24],  $p < .001$ ; autism diagnosis:  $b = 9.94$  [95% *CI*, 5.07 to 14.81],  $p < .001$ ). Autism diagnosis was linked to an average increase corresponding to 1.15 standard deviations in the SWEAA total score. Regarding specific eating problems, regression models adjusted for covariates showed that autistic traits and autism diagnosis were associated to four of the SWEAA subscales (A. Perception, D. Eating behavior, E. Mealtime surroundings, F. Social situation at mealtime), reflecting mealtime issues with sensory sensitivity, selective eating, inflexibility, and adapting behavior to others.

A significant interaction effect indicated that the link between autistic traits and overall eating problems was particularly strong in females ( $b = 0.14$  [95% *CI*, 0.07 to 0.21],  $p < .001$ ) compared to males ( $b = 0.06$  [95% *CI*, 0.01 to 0.11],  $p = .019$ ), visualized in Figure 4a. Considering specific eating problems, autistic traits and autism were associated with the E. Mealtime surroundings subscale only in females ( $b = 0.20$  [95% *CI*, 0.09 to 0.31],  $p < 0.001$ ;  $b = 10.98$  [95% *CI*, 4.25 to 17.71],  $p = .001$ ), see Figure 4b. As this subscale appeared to include questions in two differing areas, these were split into items focusing on difficulties eating in social contexts (e.g., eating in school or with friends) and non-social items (focusing on rituals and routines in mealtimes). Exploratory analyses of the two clusters revealed that the interaction effect was only found for the social context items where autistic traits and autism diagnosis was associated to more difficulties among females but not males.

In the adjusted within-pair models, the association between autistic traits and total eating problems remained when controlling for 50% of genetics in DZ pairs ( $b = 0.11$  [95% *CI*, 0.04 to 0.18],  $p = .003$ ) but not when controlling for 100% of genetics in MZ pairs ( $b = 0.04$  [95% *CI*, -0.08 to 0.17],  $p = .484$ ), and the same pattern was observed when restricting the sample to the 16 pairs categorically discordant for autism and using autism diagnosis as the exposure (DZ:  $b = 5.56$  [95% *CI*, 1.15 to 9.97],  $p = .013$ ; MZ:  $b = 2.07$  [95% *CI*, -1.25 to 5.40],  $p = .221$ ).



**Figure 4.** Association between autistic traits and SWEAA total score (a) and Mealtime surroundings subscale (b) for females and males separately. Adapted from Lundin Remnélius et al (2022).

### 4.3 Study III

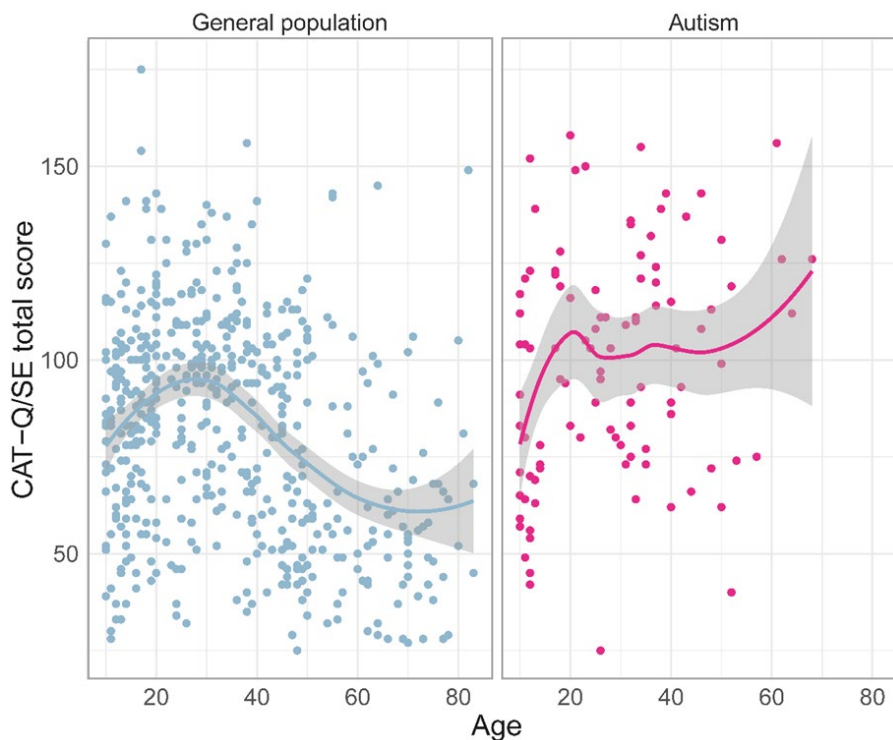
Most items performed well in the item analyses, regarding item difficulties (ranging from  $d_i = 0.40$  to  $0.69$  in the autism group and between  $0.36$  and  $0.56$  in the general population group), and item–total correlations, where most were  $\geq .30$  in the autism and general population groups, apart from items 3, 12, 16, 19, 22, and 24. Of all 25 items in the scale, 17 differentiated significantly between the autistic and the general population group.

Reliability analyses showed that the internal consistency was excellent for the total scale ( $\alpha = 0.93$ ), and good-to-excellent for the subscales (Compensation:  $\alpha = 0.92$ ; Masking:  $\alpha = 0.78$ ; Assimilation,  $\alpha = 0.85$ ). Accordingly, the CAT–Q/SE total score showed good test–retest reliability ( $r = .85$ ;  $p < .001$ ;  $ICC = .85$ ,  $p < .001$ ), and similar stability was found for the subscales (Compensation:  $r = .82$ ,  $p < .001$ ;  $ICC = .82$ ,  $p < .001$ ; Masking:  $r = .78$ ;  $p < .001$ ;  $ICC = .78$ ,  $p < .001$ ; Assimilation:  $r = .86$ ,  $p < .001$ ;  $ICC = .85$ ,  $p < .001$ ). When investigated in the subsample of participants aged 10 to 15 years, internal consistency for the total scale was excellent ( $\alpha = 0.92$ ), and test–retest reliability was moderate-to-good ( $r = .80$ ,  $p < .001$ ,  $ICC = .79$ ,  $p < .001$ ).

Supporting diagnostic validity, a main effect of group showed that autistic participants scored higher than non-autistic ( $F(1,625) = 23.54$ ,  $p < .001$ , partial  $\eta^2 = 0.04$ ), and an interaction effect was observed where autistic females scored higher than autistic

males ( $t(89.08) = 2.51, p = .014, g = 0.51$ ), whereas no difference was found in the general population group. However, the ROC-curves yielded an acceptable Area Under the Curve (*AUC*) only for participants who were 15 years or older ( $AUC = .714$ ). Regarding concurrent validity, the observed correlations between CAT-Q/SE scores and theoretically neighboring measures overall reflected the expected correlation strengths with a few exceptions. Still, the expected pattern of correlations was not fully replicated. For example, the association between CAT-Q/SE and autistic traits (AQ;  $r = .44$ ) was not stronger than observed associations with measures of social functioning difficulties (WHODAS 2.0;  $r = .52$ ), depression (PHQ-9;  $r = .48$ ), or anxiety (GAD-7;  $r = .45$ ). A similar pattern was found in the subsample aged 10 to 15 years, but the correlation with autistic traits was descriptively weaker in this group ( $r = .28$ ). Regarding factorial validity, the CFA indicated lacking fit for the original three factor structure ( $CFI = 0.828, RMSEA (90\% CI) = 0.157 (0.153-0.161), SRMR = 0.091$ ). An EFA was thus fitted, which endorsed a two-factor solution where all items except the reverse-coded loaded to factor 1 whereas the reversed items loaded to factor 2. A follow-up CFA with all reversed items removed showed improved fit indices ( $CFI = 0.961, RMSEA (90\% CI) = 0.091 (0.085-0.096), SRMR = 0.040$ ).

Scatter plots fitted with LOESS curves implied differing trajectories among autistic people and non-autistic people. A continuous decrease of self-reported CAT-Q/SE total scores was observed during adulthood in the general population sample while no corresponding decrease could be observed in the autistic sample, see Figure 5.



**Figure 5.** Scatter plots and LOESS regression curves with 95% confidence intervals visualizing the relationship between age and CAT-Q/SE total score for the general population and autism samples. (Lundin Remnélius & Bölte, 2023).

#### 4.4 Study IV

Autistic participants displayed increased camouflaging compared to non-autistic participants ( $b = 39.79$  [95% CI, 26.38 to 53.19],  $p < .001$ ), and an autism by sex interaction was found suggesting that the link was due to autistic females self-reporting elevated camouflaging levels compared to the other groups (autistic males, non-autistic females, and non-autistic males). While the covariates age and sex were not associated to camouflaging, ADHD was linked to increased camouflaging ( $b = 15.69$  [95% CI, 7.17 to 24.20],  $p < .001$ ).

Camouflaging was negatively associated with QoL across individuals, also when controlling for sex, age, autistic traits, and ADHD ( $b = -0.12$  [95% CI -0.17 to -0.07],  $p < .001$ ), where a one standard deviation increase in CAT-Q/SE total score was linked to a decrease of 0.46 standard deviations in EUROHIS-QOL score. The association between camouflaging and QoL remained in within-pair analyses in DZ and MZ pairs, implicitly controlling for shared environment as well as ~50% and 100% of genetic factors, also when adjusting for autistic traits and ADHD (DZ:  $b = -0.13$  [95% CI -0.21 to -0.06],  $p < .001$ ; MZ:  $b = -0.07$  [95% CI -0.14 to -0.01],  $p = .016$ ).

The results across individuals and within pairs did not change when internalizing psychiatric conditions was added as a covariate in the adjusted models to ensure that the link between camouflaging and QoL was not primarily underpinned by the presence of internalizing conditions.



## 5 Discussion

### 5.1 Study I

This qualitative study explored perceptions of sex/gender differences in functioning among autistic people in an international group of professionals. Thereby, the study yields a cross-cultural perspective on the topic rarely represented in autism research. The results suggests that autism professionals experience autism manifestations in females as more difficult to identify and less aligned with the common conceptualization of autism, which could be attributed to the reports of an overall less salient behavioral presentation including internalizing rather than externalizing issues, increased sociability, along with camouflaging strategies further reducing the visibility of social-communication challenges. Furthermore, the functional perspective of the study allowed us to identify environmental factors such as gender expectations and access to supportive friendships which could play important roles in outcomes for autistic females and males.

In the *Navigating the social environment* main category, autistic females were perceived as showing more social interest and a drive towards engaging in social interaction and relationships, which was reported by professionals working with children, adolescents, and adults. Social motivation has previously been hypothesized to be a risk factor for social failures and for subsequent mental health problems in autistic people (Lai & Baron-Cohen, 2015), which is corroborated by our qualitative results highlighting social motivation as a candidate risk factor for detrimental outcomes particularly among autistic females. Camouflaging was linked to female autism presentations across age groups by several professionals and was experienced even among autistic preschool girls, highlighting the need for better understanding of how such behaviors develop throughout the lifespan.

The linking of subcategories from the content analysis to ICF codes revealed that the majority (97%) of the functional areas where professionals perceived sex/gender differences were captured in the comprehensive ICF Core Set for autism, supporting the validity of this short list in covering functional areas where autistic females differ from males on the autism spectrum.

The results also revealed some discrepancies between professionals from HICs and MICs regarding the areas that were reported as differing between autistic females and males. While referenced by approximately 24% of participants from HICs, none of the participants working in MICs discussed sex/gender differences in camouflaging. These discrepancies could reflect cultural influences on behavior among autistic people, or alternatively, differences in access to diagnosis and services for autistic people between countries.

## 5.2 Study II

In this study, autistic traits and autism were linked to total eating problems and with specific eating and mealtime problems including sensory issues, selective eating, and difficulties in adapting behaviors to others. Eating problems in autism have been predominantly studied among children, but our findings are in congruence with the studies in older age groups suggesting that such issues are prevalent also among autistic adolescents and adults (Karlsson et al., 2013; Spek et al., 2020). The findings in **study II** extends previous evidence by using both categorical and dimensional measures of autism, and by controlling for potential confounding from intellectual disability, ADHD and internalizing conditions, which are common in autism and linked to eating problems (Gal et al., 2011; Karjalainen et al., 2016; Lai et al., 2019; Lyall et al., 2017).

Our results further indicate sex/gender moderation on the link between dimensional and categorical autism and eating problems. The finding that autistic traits show a relationship with overall eating problems particularly in females is novel and suggests that autistic girls and women displaying elevated autistic traits are especially at risk for issues around eating and mealtimes. In addition, the data indicate that eating with other people, e.g., in school, at work or a cafe, constitutes a specific challenge tied to female autism manifestations, which could have negative effects on social participation for this group. A recent qualitative study in a sample consisting mainly of autistic women provides hypothetical reasons for our finding, as participants described avoiding communal eating environments such as restaurants and school cafeterias due to feeling embarrassed over their own eating behaviors and due to sensory issues with noise and smells (Kinnaird et al., 2019).

The within-pair results, where the association between dimensional and categorical operationalizations of autism and total eating problems remained in DZ but not MZ pairs are consistent with a genetic influence on the link, implying that common genes may influence both traits. Still these results must be interpreted with caution as confidence intervals for the regression estimate were wide in the MZ group and overlapped with those found in DZ pairs. Importantly, a genetic influence does not entail genetic determinism (Knopik et al., 2017), meaning that these findings should not be interpreted as suggesting that interventions supporting autistic people in alleviating eating problems will not be efficient.

## 5.3 Study III

The psychometric evaluation in **study III** supported the reliability of the CAT-Q/SE and provided mixed evidence regarding the construct validity of the scale, where particularly the pattern of associations with theoretically linked measures and the factor structure indicated potential issues. While concurrent validity analyses generally yielded correlations of the expected directions and strengths, they did not fully reflect the



hypothesized pattern. For example, the associations between CAT-Q/SE scores and depression or anxiety were descriptively stronger than associations with autistic traits. Mental health problems could be consequential to camouflaging as suggested in qualitative studies (Hull, Petrides, et al., 2017), yet the strong links found in our data could also be suggestive of potential confounding from mental health problems, indicating that camouflaging may not be a response specific to elevated autistic traits but potentially to a wider range of psychopathology.

In line with our hypotheses and supporting diagnostic validity, the CAT-Q/SE total score was significantly higher in the autistic group compared to the general population group, and autistic females scored higher than autistic males. Calculating ROC-curves was of relevance as we received anecdotal examples of clinicians interpreting CAT-Q scores above a certain cut-off as indicative of autism. However, acceptable differentiation between autistic and non-autistic groups was only found among those 15 years or older, with modest sensitivity and specificity, suggesting that the reported cut-off for the total score should not be used to make diagnostic judgements even in this group.

Our psychometric evaluation pointed towards potential issues with the reverse-coded items in CAT-Q/SE. The item-level analyses showed that six items were less consistent with the total scale overall, and among these all five reverse-coded items were included. The reversed items also loaded to their own factor in the EFA suggesting that participants responded to this cluster of items in a certain way, which could be indicative of confounding from other factors than camouflaging (Suárez Álvarez et al., 2018). The fit was improved in a second CFA where the five reversed items were excluded suggesting that further evaluation of a revised version of the CAT-Q/SE without the reversed items or where these items are reformulated to positive wordings could be fruitful. Overall, the psychometric evaluation provided in **study III** give preliminary support for using CAT-Q/SE as a measure of camouflaging in Sweden, while simultaneously pointing towards issues regarding the construct validity of the scale which should be further investigated.

Furthermore, our findings provide insights regarding the construct of camouflaging and elucidate aspects that are likely largely overlapping between autistic and non-autistic populations. Item validity analyses showed that most items in the CAT-Q/SE differentiated between autistic and non-autistic people, but also indicated behaviors that seem to be as common in both groups. For example, these include experiences of attempting to appear relaxed in social situations and constantly attending to the impression made on others. On the other hand, items that did differentiate between the groups likely reflect more autism-specific aspects, including experiences of “performing” rather being oneself in social interaction and trying to learn how others use non-verbal communication from television or literature.

Our study is the first to explore the relationship between age and camouflaging across a wide age range, where the LOESS trend curves imply that such behavior may develop differentially in autistic and non-autistic people. While camouflaging may overlap with impression management in the general population, our findings suggest that these behaviors may be less important in the lives of non-autistic people as they get older, whereas behaviors aiming to mask or compensate for autistic traits are used extensively throughout adulthood for many autistic people. This finding could reflect that similar behaviors are used more voluntarily by neurotypical people to present their most favorable characteristics as has been suggested (Ai et al., 2022). Declining use of such behaviors may then be due to less concern about the impression made on others as many experience being more settled in their private and professional lives. In contrast, autistic people may not experience a similar reduction in the external pressures, such as autism-related stigma, that underpin camouflaging in this group (Hull, Petrides, et al., 2017; Perry et al., 2022).

#### **5.4 Study IV**

For **study IV**, the initial plan was to use a discrepancy approach to measure camouflaging, similar to the operationalization in previous studies (Lai et al., 2017). However, after consideration of the potential methodological limitations of discrepancy measures (Furr, 2011), a decision was made to use self-reports on the CAT-Q/SE instead. As the questionnaire was not included from the beginning of the RATSS project, efforts were made to collect CAT-Q/SE data from twin pairs who already had participated in RATSS via a web survey.

Among the twins in **study IV**, autistic females were found to camouflage autistic traits more extensively than autistic males and non-autistic participants, in line with reports in some but not all previous studies (Cook et al., 2021). Still, the groups were small in our sample (eight autistic females and 14 autistic males). In addition to autism, ADHD was also linked to increased camouflaging, potentially suggesting that the camouflaging operationalization in the CAT-Q also pick up behavioral strategies found across neurodivergent populations and psychiatric conditions rather than being specific to autism.

The findings in this study confirm and extend the results of previous studies, including **study III**, reporting negative correlations between camouflaging and wellbeing. The within-pair analyses indicate that the association is not due to familial confounding. Thus, the co-twin control design allows us to falsify a range of scenarios where the phenotypic link is due to familial confounding such as pleiotropic effects of genes and permits a stronger causal claim than previous cross-sectional studies. This is an important step towards identifying risk factors for low QoL in autistic and non-autistic

populations, which may be of particular importance for autistic people and even more so in autistic females, who display elevated camouflaging.

## 5.5 Limitations

A limitation of this thesis is the predominant focus on female and male sex/gender. Although the survey question in **study I** asked about gender differences in general rather than differences between females and males, and **study III** included participants identifying as non-binary, these studies nevertheless mainly yielded data regarding females and males. Future studies should include large and gender diverse samples to allow investigations in the variation of gender identities encompassed in the autism population.

The results in **study I** need to be interpreted considering the large proportion of the total 225 experts who did not respond to the question concerning sex/gender in autism ( $n = 91$ ) or explicitly responded *not* perceiving differences between autistic females and males ( $n = 32$ ). The cause of the lacking responses can only be speculated about as comparisons of demographic variables between groups yielded no significant differences. Nevertheless, the lacking and “no” responses indicate that a proportion of autism professionals do not perceive functional sex/gender differences in autism. Speculatively, other factors such as low awareness of sex/gender-related factors in autism, limited experiences of autistic females, and ascertainment bias with mainly autistic females showing autism manifestations similar to those common in males being diagnosed could reduce the likelihood of professionals reporting differences. The phrasing of the question could also have played a role, in asking for aspects “specific” to gender, which could have averted reports of more subtle differences. Second, while the sample represented both HICs and MICs, there was a lower representation of professionals from MICs potentially limiting the perspectives provided in our results. In addition, professionals working in low-income countries were not included in the qualitative analysis, reflecting difficulties in identifying autism professionals from low-income countries in the recruitment process. Thus, the generalizability of our findings to these countries is uncertain. Third, no respondent validation of the content analysis was conducted. However, to promote credibility multiple analysts were involved and the two co-authors not initially involved in the analysis conducted audits of the categories and included qualitative data (Elliott et al., 1999). Fourth, the use of a single survey question likely increased the risk of misinterpretation and ambiguous responses and limited the depth of the qualitative data collected. However, the use of an email survey allowed collection of information from a broad international and multidisciplinary pool of experts providing rare perspectives to the field of sex/gender differences in autism. Fifth, the requirement to respond in English may have influenced the richness of data as a large proportion of the participants did not have English as their first language. Finally, the data collection was conducted in 2013 and 2014, and perspectives among professionals

may have shifted slightly since then, following increased awareness and research interest in the topic of sex/gender in autism.

Some limitations are applicable to both **study II and IV**. First, the RATSS sample is not randomly selected and findings from the across-individuals analyses may thus not extend to the general population. Similarly, a common criticism of twin research concerns the generalizability of findings in twins to non-twin populations, particularly since the elevated risk of some prenatal events such as low birth weight could potentially increase the likelihood of autism in this group. Still, research in population-based samples have not found higher rates of autism or higher levels of autistic traits in twins compared to singletons (Curran et al., 2011; Lundström et al., 2015). A second limitation is the reduction of statistical power in the within-pair analyses, where the total sample is split in DZ and MZ pairs and only twin pairs that are discordant for the exposure or the outcome will contribute to the regression estimate (Allison, 2009). In **Study II**, only two pairs (both MZ) displayed no intra-pair variation in either exposure or outcome, and in **study IV**, intra-pair variation in either exposure or outcome was observed in all twin pairs, suggesting that most of the twin pairs were informative for the analyses in both studies. In addition, within-pair associations are particularly susceptible to measurement error, and this is especially pronounced within MZ pairs, which can attenuate the associations found in these analyses (McGue et al., 2010). On balance, even though the sample size is limited in these studies, the RATSS twin sample is phenotyped through a comprehensive assessment procedure providing a depth of data which is not commonly reflected in larger twin studies. The findings in RATSS can thus extend findings in larger twin studies, and vice versa.

In **study III**, autism diagnosis was self-reported along with age of diagnosis and was not validated further. Still, autism diagnoses reported in web-based studies have been found to be largely valid, for example when verifying parent-reported autism diagnosis with clinical records (Daniels et al., 2012). A second limitation is that the survey included measures that have not been validated in children. We mainly found comparable associations between CAT-Q/SE scores and these measures in children between 10 and 15 years and the subgroup aged 15 and older, implying that participants in the younger age groups were able to provide accurate reports. Nevertheless, this limitation needs to be considered when interpreting the results in this subgroup. Third, the lower CAT-Q/SE total score among autistic participants aged 10 to 15 years must be interpreted in light of this subgroup being relatively small ( $n = 28$ ) and including proportionally fewer females compared to the subgroup of autistic participants aged 15 years or older. Fourth, the sample included eight participants self-reporting intellectual disability, and the original CAT-Q was not validated in this group. These participants were included as their self-ratings were not notably different from others. Nevertheless, in the process of collecting data for **Study IV** some difficulties in interpreting the CAT-Q/SE items were

noted among participants displaying low verbal ability, suggesting that the utility of the CAT-Q/SE among people with intellectual disability is limited.

Both **study III and IV** relied on self-report measures, including the key constructs of camouflaging and QoL, requiring that participants have a level of insight and awareness in order to provide valid ratings. This entails that we may not have captured camouflaging in people who are not aware that they are using such behaviors (Pearson & Rose, 2021). On the other hand, the first-hand perspective is essential for these constructs. For example, QoL is defined by the individual's subjective appraisal of their own position in life (World Health Organization, 1998). Correspondingly, other people may not be able to observe the camouflaging attempts or intentions that can be captured by the CAT-Q. Furthermore, comparisons of self-reports and proxy-reports show that the two perspectives are not interchangeable for neither camouflaging nor QoL (Hannon et al., 2022; Knuppel et al., 2018; Moss et al., 2017). Additionally, while the operationalization of QoL used in **Study III and IV** allow people to report their subjective appraisal of wellbeing, use of general population measures of QoL in autistic populations has been questioned as such measures may not pick up on facets of QoL that are particularly salient for autistic people (McConachie et al., 2018; Moss et al., 2017), meaning that the measure used may have missed aspects of QoL especially important for the autistic participants in these studies.

For **study IV**, the construct validity issues concerning the CAT-Q/SE reported in **study III** constitute a limitation, including questions regarding diagnostic validity among autistic children and the general issues with the reversed items in the scale, along with the difficulties in replicating the factor structure reported for the original CAT-Q. Second, for most of the RATSS twins included in **study IV**, the diagnostic assessments of autism and internalizing conditions and the collection of parent-reported autistic traits on the SRS-2 were made at an earlier time point compared to when the main variables camouflaging and QoL were collected. While both autistic traits and internalizing conditions and symptoms show reasonable stability over time (Carballo et al., 2010; Constantino et al., 2009; Haraguchi et al., 2019; Nivard et al., 2015; Robinson et al., 2011), the data in **study IV** may not represent precise measures of the participants' current status regarding these variables.



## 6 Conclusions

This thesis examined female autism phenotypes and sex/gender differences in autism. The results point toward sex/gender moderation in various areas which could contribute to the risks of delayed diagnosis and detrimental outcomes especially pronounced in autistic females.

Qualitative data involving the perceptions of autism professionals implicated areas which should be acknowledged to enhance timely recognition of autism in females. Female phenotypes were seen as more difficult to recognize and less aligned with the conceptualization of autism, suggesting that increased awareness of behavioral examples common in autistic females may support identification of female manifestations. Such examples were suggested in the qualitative data, including restrictive interests in “female-typical” topics, increased motivation to socialize and develop relationships in combination with social-communication challenges, and camouflaging strategies including practicing non-verbal communication. In addition, environmental factors were identified, including gender-related expectations of increased social competence in girls, and access to supportive friendships, which could have an impact on functioning, wellbeing, and mental health outcomes in autistic females.

The results further support sex/gender moderation on the link between autistic traits and overall eating problems, which was found to be particularly strong in females. A female-specific association was also found between autism and difficulties with eating together with others in social settings, which could limit social participation opportunities for autistic females. These findings require replication, but nevertheless highlight the importance of professionals in health care services being aware of eating problems in autistic adolescents and adults and that such issues may require assessment and subsequent management, especially when encountering females with elevated levels of autistic traits.

This thesis supports the hypothesis of a link between camouflaging and female autism manifestations, while acknowledging that some autistic males also use such behaviors to mask or compensate for autistic traits extensively. Furthermore, the studies elucidate some aspects of the construct. First, while camouflaging behaviors are closely associated to autism, especially among those identifying as female or assigned female sex at birth, these strategies may be common also in a broader group of people displaying psychopathology or neurodiversity, including elevated symptoms of depression and anxiety, general challenges in social functioning, or ADHD. Some camouflaging behaviors were found to be equally common among autistic people and people in the general population, suggesting that aspects of the construct reflect universal human attempts to manage the impression of others. Still, while camouflaging

behaviors appear to decline steadily with higher age among non-autistic people such strategies remained elevated throughout adulthood for autistic people, potentially reflecting different motivations for camouflaging in the two populations. Furthermore, the findings in this thesis strengthen previous claims of a causal impact of camouflaging on QoL, where a negative link between these two constructs remained also when controlling for a variety of alternative non-causal explanations in a co-twin control design. These results highlight the need for further research shedding light on how similar strategies are used across clinical and neurodiverse groups, as well as longitudinal research which could further investigate the detrimental consequences of camouflaging that was supported in this thesis. In addition, the findings endorse the development of supportive interventions in alleviating camouflaging which likely will require that both individual and environmental factors are addressed. Such interventions could be fruitful in supporting QoL and prevent mental health problems especially among autistic females.



## 7 Points of perspective

In **study I**, autism professionals experienced female autism phenotypes as more difficult to recognize as autistic and less corresponding with the criteria in diagnostic manuals. Potentially, this could be mitigated with increased awareness of manifestations of autistic characteristics more common in females, including those reported in the study such as social motivation in combination with social-communication challenges, and camouflaging. The results also suggest that gender-related environmental factors influence functional outcomes in autism which deserves follow-up in future research. Such investigations could utilize the functional perspective provided by the ICF, allowing self-, parent-, and clinician-reported data to be linked to ICF codes and thus harmonized so that data collected from different sources can be compared. Acknowledging environmental factors along with strengths and limitations pertaining to the individual will allow more holistic investigation of person-environment fit among autistic females and males, identifying mismatches between environmental demands and individual abilities that may have negative consequences for functioning, mental health, and QoL (Lai & Baron-Cohen, 2015; Mandy, 2022). Such research should acknowledge the hindering and helpful factors moderated by sex/gender reported in our findings, including areas such as access to friends, access to health care, and gendered social expectations along with their interaction with social motivation and camouflaging. In addition, the discrepancies suggested by the explorative comparisons between HICs and MICs emphasize that more autism research, including in sex/gender differences, needs to be conducted in low- and middle-income countries where the majority of autistic people live, to gain insights into how generalizable findings from HICs are (Hahler & Elsabbagh, 2015).

In **study II**, the sex/gender moderation on the link between dimensional and categorical operationalizations of autism and eating problems correspond to preliminary evidence in indicating that females displaying elevated autistic traits are especially at risk for a range of eating problems. This sex/gender influence needs replication in larger non-selected samples. Still, our results warrant further investigations into the potential impacts of eating problems on social participation and QoL in autistic adolescents and adults. Future research should also study the link between autism and eating problems in population-based twin samples using cross-twin cross-trait correlations allowing quantification of the degree of genetic, shared environmental, and NSE influence on the link.

Currently, eating problems in autistic adolescents and adults are rarely assessed, yet our findings indicate that this area deserves attention in health care services. These issues may not take the form of disordered eating behaviors associated with AN or bulimia nervosa but may nevertheless impact the lives of autistic individuals. In addition,

recent research point towards eating problems common in autism, e.g., selective eating, as a potential target for hindering development of later eating disorders (Carter Leno et al., 2022), further highlighting that common eating problems in autistic adolescents and adults should be addressed by health care. Furthermore, given that autistic girls and women with high levels of autistic traits struggled with eating issues in general as well as social eating problems, addressing these issues may be of particular importance in this group.

This thesis provides broad support for a link between female autism manifestations and camouflaging, which was endorsed by professionals' qualitative accounts in **study I**, as well as quantitative analyses based on gender identity and self-reported diagnosis in **study III**, and sex assigned at birth and best-estimate clinical consensus diagnosis in **study IV**. Furthermore, **study III and IV** provided novel information regarding the construct of camouflaging warranting future elaboration. The item-level findings in **study III** point towards aspects of camouflaging that are more characteristic of autism as well as aspects showing no differentiation with the general population. Future studies should further explore camouflaging levels across psychiatric and neurodivergent populations, further endorsed by the increased camouflaging in ADHD found in **study IV**. Given the questions raised regarding construct validity in **study III**, further psychometric evaluation of the CAT-Q/SE is warranted, including testing if rephrasing or removing reversed questions improves the psychometric characteristics of the questionnaire.

In **study IV**, the findings of a robust association not explained by familial confounding support and strengthen the claim of a causal effect of camouflaging on QoL. Future studies should employ family-designs in larger samples in order to replicate our results, as well as longitudinal designs to disentangle the direction of the association. Also, the results endorse the development of interventions supporting people who camouflage extensively, including neurodivergent people and particularly autistic females, to reduce unhelpful camouflaging. Any support needs to acknowledge the complexities of camouflaging as such strategies may be experienced both as detrimental and helpful (Hull, Petrides, et al., 2017). Additionally, interventions likely need to target environmental factors in order to be fruitful given that camouflaging in autism is often a reaction to autism-related stigma and external pressures (Cage & Troxell-Whitman, 2019; Perry et al., 2022). Furthermore, QoL has a protective effect on suicidality (Chioqueta & Stiles, 2007), implying that support in reducing camouflaging could simultaneously facilitate overall wellbeing and mitigate the risk of premature mortality in autistic people.

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