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### Autism as a multicausal system

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**Publication date**

2019

**Document Version**

Other version

**License**

Other

[Link to publication](#)

**Citation for published version (APA):**

Deserno, M. K. (2019). *Autism as a multicausal system*.

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METAPHORS WE THINK BY

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Traditionally, the stylistic device of the metaphor — a reference to an object or action in terms of something similar — belongs to the realm of rhetoric, which is a definitive feature of disciplines such as jurisdiction and literature. Within any scientific discourse, however, people often engage in the use of well-established metaphors to translate abstract relational or causal hypotheses into intuitive examples. In the process of theory formation, experts systematically use inference patterns from one conceptual domain to reason about another. From studying such dominant metaphors, one can learn a great deal about the field-specific *Zeitgeist* of theoretical reasoning.

Metaphors for human complexity, for example, have a long history in drawing from mechanistic phenomena: the brain is wired like a computer, the body resembles a clockwork and our psychological well-being should be thought of in terms of charged or drained batteries. Thinking alongside these mechanistic metaphors has enabled medical doctors and researchers to establish a highly successful account of the relation between what can be seen, i.e. symptoms, and the underlying, or hidden, cause of these observables. This relational account is a fundamental pillar of the diagnostic process in the medical sciences: doctors can link yellowish skin to a dysfunctional liver, or coughing and sneezing to the presence of a virus. Along the same line of thought, it is often assumed that co-occurring psychiatric problems stem from a common cause within individuals (Borsboom et al., 2017). Mechanistic thinking about the human mind and body has thus resulted in the reductionist assumption that a present set of symptoms can be linked to some traceable etiological agent. These metaphors, however, are usually highly time-dependent and change alongside empirical advances and technological innovation (Lakoff & Johnson, 1980). They nonetheless shape a great deal of how we study the human mind in its so-called ‘typical’ and ‘atypical’ variants.

Now, the twenty-first century has witnessed a shift away from reductionist-inspired mechanistic metaphors towards a more complexity-aspiring metaphorical framework (Barabasi, 2012). One prominent example is the popular ecosystem metaphor for human complexity in which complex higher-order phenomena, such as intelligence or psychopathology, emerge from the interplay of genetic, neural and behavioral factors. What follows from this is that one can conceptualize heterogeneity in human development as the result of the many different outputs a complex biodiverse ecosystem can have (Van de Leemput et al., 2014). This metaphorical framework has in-

spired a relatively young branch of psychological research to focus on complex interrelations, or networks, of psychological attributes to accommodate the complex dynamics of the human mind (Van der Maas et al., 2006; Cramer et al., 2010; Borsboom & Cramer, 2013).

### 1.1 AUTISM

Recently, the metaphor of a complex ecosystem has also found its way into the field of (a)typical development. More and more researchers argue that here might not be one deterministic cause but a variety of causes that merely change the probability of an impairment to emerge (Johnson, 2017; Kievit et al., 2017; Van der Maas et al., 2006; Kendler et al., 2011). Essentially, atypically developing, autistic<sup>1</sup> individuals are diagnosed based on a mechanistic account of the co-occurrence of three characteristics: social impairment, communicative impairments and repetitive motor and restricted interests (American Psychiatric Association, 2013). It is important to note that these diagnostic criteria make autism a *behaviorally* defined condition.

Along the lines of the above-mentioned reductionist idea, it has long been thought that these characteristics stem from one common cause, a sometimes implicit assumption that still motivates various researchers in the autism field to keep looking for a single biomarker (Anderson, 2015). Although multiple potential risk factors have been identified, a replicable hierarchical cascade of events leading to an autistic profile has not yet been found (Johnson, 2017). Simultaneously, the field is slowly moving away from the assumption that all autistic individuals share one particular etiological agent.

A popular theoretical model by Happé and colleagues (2006), for example, argues that the three core characteristics of autism are probably caused by distinct deficits that can occur in isolation. The often observed intra-individual co-occurrence of these deficits, however, remains difficult to explain from such a theoretical stance (Brunsdon & Happé, 2014). A growing body of literature is now picking up on a more complexity-aspiring framework and toolbox to study atypical development, its antecedents, risk factors, and outcome (Johnson, 2017; Anderson, 2015). This theoretical shift leaves us with a new focus on multicausal, multivariate patterns that span several levels of interest (e.g., environment, behavior, neuropsychology etc.) and forms the methodological and theoretical basis of this thesis. With this novel focus we seek to investigate the two biggest issues in the atypical development realm, and autism specifically: the development of autistic

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<sup>1</sup> The appropriate terminology used to describe autism is a topic of recent debate (Kenny et al., 2015) and remains a matter of personal opinion. In this thesis, we use person-first (e.g., individual with autism) and identity-first language (e.g., autistic individuals) interchangeably to refer to people with an autism diagnosis.

characteristics and the generally low quality of life (QoL) of this population.

## 1.2 DYNAMICS IN OUTCOME

In the past decades, many studies have reported on a variety of outcome variables for the autism population. Traditionally, the focus has been on percentages of individuals who live independently, work for their own income, or engage in long-term relationships. Focusing on these outcome parameters has resulted in the assumption that autistic individuals show a relatively low outcome (Kuhltau et al., 2010; Billstedt et al., 2005; Howlin et al., 2004), also in comparison to other clinical populations. Discussions concerned with what objective and subjective parameters to include in a proper evaluation of a 'good life' date back to ancient Greek philosophers such as Aristotle (see Franklin (2010) for a historical perspective on the definition of a good life). With our scientific thinking shifting towards more multivariate conceptualizations, however, we can now evaluate the direct interplay between those traditional, or 'objective', parameters of QoL versus someone's subjective evaluation of their QoL (Constant et al., 2018; Stokes et al., 2017; Begeer et al., 2017; Van Heijst & Geurts, 2015).

The call for this shift of focus stems from the finding that well-researched predictors of better outcome in autism, such as autism severity (Eaves & Ho, 2008), IQ and language development, do not seem to be related to QoL in a replicable and straight-forward pattern (Van Heijst & Geurts, 2015; Kuhltau et al., 2013; Renty & Roeyers, 2006). It has, therefore, been argued that extending traditionally assessed outcome variables with self-reported subjective well-being would do justice to the complex patterns of interrelated factors that together constitute an individual's QoL. The theoretical shift towards conceptualizing autism as an adaptive developmental pathway led to a growing focus on investigating multivariate patterns surrounding atypical developmental outcomes, which can be seen as the second building block of this PhD thesis.

In Chapter 2, we created the first network of interrelations among subjective evaluations of QoL in adults with autism to learn that there are differentially related direct predictors of the general life satisfaction of adults with autism. Based on this first orientational study, we conducted a second study, reported in Chapter 3, combining self-reported subjective well-being measures with measures of (objectively defined) good outcome in adults with clinically identified autism. We were interested to see what aspects of autism symptomatology do (directly) relate to aspects of more subjectively defined QoL measures (e.g., general life satisfaction) versus more objectively defined measures of functioning (e.g. someone's living situation). In addition, we aimed to illustrate how representing psychological variables

as higher-order (i.e. sum-scores) or lower-order (i.e. sub-scales) constructs can clarify their functional interdependencies on different levels. Discussions about these different construct levels of psychological phenomena led to a collaborative methodological project, reported on in Chapter 4, in which we proposed community detection as a means to study the role of specific elements in multivariate patterns and their organization into communities. This novel focus on the community level of psychological network structures offers a framework to study essential clinical concepts such as comorbidity, heterogeneity and hallmark symptoms.

By exploring the potential of this novel theoretical and statistical approach to investigate multivariate systems (such as QoL in autism), we laid the groundwork for the projects to follow: digging deeper into the field of QoL in autism, its bottlenecks became exceedingly clear as well. First, there is an ongoing discussion about the respective incongruence between self- and proxy-reported assessments of QoL in individuals with autism. Second, there is a persistent lack of large-scale longitudinal data providing insights on factors relevant to developmental trajectories of QoL in autism. We address both issues in two subsequent projects: in Chapter 5 we took the factors included in the network structure in Chapter 3 and constructed a symptom network on the basis of clinicians' judgments (Perceived Causal Relation methodology; Frewen et al., 2012). In a next step, we looked at the congruency of clinicians' rated causal relationships among characteristics of autism and well-being with the network structure obtained from statistical analyses of the survey data in Chapter 3. In Chapter 6 we took a data-driven approach to uncovering subtypes in multivariate patterns associated with longitudinal changes in QoL. The analysis reported on in this chapter is executed on unique longitudinal data from the Netherlands Autism Register (NAR).

### 1.3 DYNAMICS IN DEVELOPMENT

The final project of this thesis arose from the synergy of complexity-inspired theoretical advances in the developmental field and unique developmental data on children 'at risk' for atypical development, and autism specifically. With autism being behaviorally defined, we propose that the field can benefit from the developmental branch of the network approach to psychological constructs. Within this branch, the *mutualism model* is most prominent, introducing a network of multiple interacting and mutually reinforcing traits and factors to describe human development (Van der Maas, 2006). This model has originally been developed to explain the positive manifold of correlations seen for cognitive processes contributing to IQ (Van der Maas et al., 2006). More recently, it has also been suggested that the mutualism model might offer a fruitful theoretical framework for the

'multifinality' concept in developmental psychology, and more specifically autism (Anderson, 2015). Based on this model, one would hypothesize that the observed heterogeneity in autism should be interpreted as a result of the potential for small change to produce large non-linear effects on emergent phenotypes, similar to the heterogeneity we observe in typically developing individuals. Combining the lessons learned from previous projects, where we showed the importance of zooming in on a detailed description of functional interdependencies, the mutualism model offers a framework to investigate the role of specific parameters for longitudinal dynamics in atypical development. In Chapter 7 we, therefore, adopt a longitudinal approach to investigate how different characteristics of impairment co-develop, informing us on dynamic processes that drive atypical development. Eventually, such informed longitudinal models could enable us to detect developmental challenges in an early stage and intervene before they self-reinforce over time. Finally, Chapter 8 contains an overview of the results of this thesis, accompanied by a general discussion of possible future endeavours based on the presented results and theoretical framework.