

# To each their own

### Developmental trajectories in autism

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In typically developing children, developmental trajectories across different domains are very well defined. This is not the case in autism. During early childhood, typically developing children go through periods of accelerated development, according to an established timeline. Whether it be social, language, physical or cognitive skills, we usually observe these acquisitions in the same order and at approximately the same age in all children. In autistic children, some of these steps are delayed.<sup>1</sup> These delays, such as a lack of verbal language development or lack of response to their name being called, are usually what lead parents to seek medical advice. A great deal of variability, or heterogeneity, exists across the developmental trajectories of children who go on to receive an autism diagnosis. Researchers took a closer look at this variability and sought to characterise it, to better understand heterogeneity in autism.

### What was their study?

This study was conducted by Susan Kuo and her US colleague in JAMA *pediatrics*, a prestigious research journal. Kuo's research merged the results of 17 098 autistic children across four large research cohorts. The research team's goal was to describe the ages at which autistic children reached certain developmental milestones, such as : smiling, sitting, crawling, walking,

spoon feeding, first words, first sentences, and potty training. The authors then tried to explain the differences, otherwise known as variability, in when children reached these steps.

### Genetics or environment?

The authors firstly demonstrated that autistic children presented with developmental delays when compared to their siblings who did not have autism or an intellectual disability. This does not mean that every autistic child observed individually will present with a delay compared to their sibling on each of the 8 milestones studied. However, the average age at which the autistic group reached each milestone was significantly higher than the average age at which their siblings with no diagnosis reached the same step (sibling development was comparable to results found in the general population). Delays observed in autistic children are therefore highly linked to diagnosis, as opposed to family environment or family genetic predispositions. The authors then tried to identify the characteristics which could explain different developmental trajectories within the autistic population.

## How can we explain difference in developmental trajectories?

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- The term « delay » refers to a significant difference between the age at which a milestone is reached and the age at which this is usually observed in typically developing children. We could also say that this took longer.
- 2 These genetic conditions are often associated with intellectual disabilities.

## spectre



### Study results in autism research are highly dependent on diagnostic practices and who gets included in samples.

#### Original article:

Kuo, S. S., van der Merwe, C., Fu, J. M., Carey, C. E., Talkowski, M. E., Bishop, S. L., & Robinson, E. B. (2022). Developmental variability in autism across 17 000 autistic individuals and 4000 siblings without an autism diagnosis: comparisons by cohort, intellectual disability, genetic etiology, and age at diagnosis. JAMA pediatrics, 176(9), 915–923. doi:10.1001/jamapediatrics.2022.2423 For autistic children with a genetic condition, expected developmental delays closely mirrored those of autistic children with an intellectual disability, since these conditions are usually associated with lower IQ.<sup>2</sup> The only exception was in the development of the smiling reflex, which tended to emerge at the same time in autistic children with or without an associated genetic condition.

The authors also found a link between age of diagnosis of autism and the presence and significance of developmental delays. Developmental delays were most significant in children diagnosed before age 5, followed by children diagnosed between the ages of 5 and 9, followed by children diagnosed at age 10 or older. This link does not mean that early diagnosis causes developmental delays, but rather that these two variables vary together. It would instead seem plausible that significant developmental delays would lead to early diagnosis. Indeed, children diagnosed before age 5 were more likely to have a double diagnosis of autism and intellectual disability. In comparison, children diagnosed after 5 were more likely to have only a diagnosis of autism.

Lastly, the authors compared the four cohorts (Autism Genetics Research Exchange, Autism Simplex Collection, Simons Simplex Collection et Simons Foundation Powering Autism Research Collection) which together provide data on 17,098 children. Interestingly, they found differences in the development of autistic children across cohorts. The older the cohort the children belonged to, the greater the variability of ages at which milestones had been reached. Children from older cohorts displayed greater delays in speaking their first words, first sentences and potty training than children from more recent cohorts. This finding could result from broadening diagnostic criteria over the years, as more autistic people with no intellectual disability and no speech delays (formerly Asperger's syndrome) have come to be included in the spectrum. Thus, children from more recent cohorts display less developmental milestone delays.

#### What are the main study takeaways?

Kuo and her collaborators point out that no matter the sample size, study results in autism research are highly dependent on diagnostic practices and who gets included in samples. Secondly, the presence or absence of an associated condition (ex: genetic syndrome or intellectual disability) has a significant impact on the age at which children reach developmental milestones. In sum, the large heterogeneity observed in autism research cohorts confirms the importance of using well-defined samples before making any conclusions in autism research.