

NEWS

# Autism's relationship to head size, explained

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Some people with autism have an unusually large head: This fact has been known since autism was first **described in the 1940s**. But debate about this finding has raged ever since. How many people with autism have a large head? What causes the enlargement? And does it have any bearing on outcome?

Here is what researchers do and do not know about head size in autism.

## What proportion of people with autism have a large head?

When **Leo Kanner** first described 11 children with autism in a 1943 paper, he noted many unusual features. "Five had relatively large heads," he reported, and he said no more on the matter. But the sample size was small.

Many other scientists noted the same link over the following decades. A 1999 review estimated that **20 percent of people with autism** have statistically large head size, or 'macrocephaly'<sup>1</sup>.

In 2011, the **Autism Phenome Project** refined this estimate to **15 percent of autistic boys**<sup>2</sup>. The team followed boys with autism from their diagnosis throughout childhood. They focused on whether head size is disproportionate to the rest of the body, rather than simply large. The researchers call this 'disproportionate megalencephaly' and say it marks a distinct subgroup of autistic people. "We've defined a big-brain form of autism," says lead investigator **David Amaral**,

distinguished professor of psychiatry and behavioral sciences at the University of California, Davis MIND Institute.

No one contests the 15 percent figure, but scientists differ in their interpretation of the finding.

“It only applies to a small proportion of children with autism,” says **Katarzyna Chawarska**, Emily Fraser Beede Professor of Child Psychiatry at Yale University.

Neuroscientist **Eric Courchesne** of the University of California, San Diego argues instead that unusually large brains are a near-universal feature in autistic people, and the 15 percent represent the most extreme cases of this trait. He points to a 2015 review of more than 8,000 people, which concluded that head size is larger in autistic people than in their typical peers<sup>3</sup>.

### **What about girls on the spectrum?**

Multiple studies have suggested that large brains are much rarer among **girls with autism** than among boys with the condition.

“We found overgrowth in terms of head [size] in boys, but definitely not in girls,” Chawarska says.

Earlier studies did not include many girls, so when Amaral and his colleagues set out to study the phenomenon, they preferentially recruited autistic girls. “Even though we have a much larger number now, we still don’t see it in the same frequency with girls as we do with boys,” says Amaral. “It’s still very, very rare.”

The reason for this sex difference is unclear, but autism is thought to affect girls differently than boys, with girls being somehow **protected from the condition**.

### **Do autistic children who have a large head also have a large brain?**

Yes. Researchers have scanned the brains of autistic people by using technologies such as magnetic resonance imaging (MRI) and have found that those with a large head also tend to have an unusually large brain. However, the link between the two is not entirely straightforward — some autistic **children with an enlarged brain** don’t have a large head — so it is best for researchers to scan the brain rather than rely on head measurements.

### **Which parts of the brain are enlarged?**

There is a complicated pattern of differences throughout the brain.

Much of the volume of the human brain is contained in its thick outer layer: the cortex. This seems to be where most of the enlargement happens<sup>4</sup>. “The basic organization of the cortex is not different, but there is more of it,” Amaral says.

However, it is unclear what the extra material is, because MRI scans cannot resolve those details.

The region could have **extra neurons** or support cells, or even excess cerebrospinal fluid<sup>5</sup>.

Other regions of the brain known to be associated with autism may also be enlarged. These include the fusiform gyrus, a brain region involved in processing facial information; and the primary visual cortex, which detects basic visual information. The latter may not seem like an obvious region of interest in autism, but in fact **visual problems are common** in people with the condition.

### **When in life is the enlargement present?**

The brain enlargement begins early in life. Courchesne and his colleagues found in 2003 that the brains of children with autism grow unusually quickly during their first year of life<sup>6</sup>. A 2018 study then reported that the overgrowth is detectable in utero<sup>7</sup>.

Researchers disagree on how long the enlargement lasts, however. For example, Courchesne argues that it disappears later in life, as autistic adults have an average-sized brain. Amaral's team is tracking head and brain size in autistic boys over time. They reported in 2016 that brain overgrowth persists until 5 years of age<sup>8</sup>. Amaral says his team has unpublished data indicating that the pattern continues until age 11, and he expects it to persist into adulthood.

### **Does macrocephaly have a bearing on outcome?**

Having a big brain is generally considered a good thing, but autistic children who have an enlarged brain paradoxically have a poorer outcome than those with an average-sized brain<sup>9</sup>. This finding is largely undisputed.

Large head size in the first two years of life is a good predictor of the severity of a child's autism traits at age 4. Large-brained autistic children struggle with everyday skills such as using cutlery to eat, and their skills tend to decline over the first six years of life. Macrocephaly is also associated with social difficulties and delayed onset of language<sup>10</sup>.

### **Do some people with autism have an unusually small head instead?**

Yes, some autistic people have a small head, or microcephaly, although this seems to be much less common than macrocephaly — and much less is known about it<sup>1</sup>.

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