

VIEWPOINT

Seeing connections between autism and blindness

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The ability to see plays a large role in the development of the brain — so it makes sense that there would be a connection between vision and autism, which is essentially a condition of brain development.

When the eyes are open, vision is the dominant sense. Continuous input from the eyes links stimulation from other senses into a coherent whole.

A typical newborn already looks preferentially at faces, driven by an innate motivation to interact with others¹. In this way, vision drives social-communication skills. Visual input shapes the brain during the first year of life. The visual system also processes aspects of nonverbal communication, such as shared visual attention, facial expressions, gestures and body postures.

Sight is also crucial for acquiring some basic concepts — such as cause-and-effect relationships among actions, self as separate from others and object permanence.

So it stands to reason that without vision, brain development would go awry.

As a child neurologist, I treat children with various developmental disabilities, including autism. Every year, I see at least one blind child who shows the full clinical manifestations of autism. Because congenital blindness is rare, this was striking to me, particularly because the autism traits

in these children are typically severe.

In 2012, I decided to investigate the relationship between autism and blindness in an unbiased population. Although the study was small, it indicated that autism is more than 30 times as common in blind people as in sighted people². Other work from my team suggests this relationship is specific to vision: Hearing impairment is not strongly connected to autism³. The association is also independent of intellectual ability, showing that problems with cognition alone cannot explain the connection⁴.

Blind science:

The first report of autism in blind children appeared in more than 60 years ago⁵. In that study, the researcher identified autism in 5 of 60 infants who became blind because of retinopathy of prematurity, a condition in which the retina does not fully develop. (The rest of the children also had mild autism traits.)

Since then, scattered reports have drawn an association between congenital blindness and autism traits. But professionals typically did not recognize children's social and other difficulties as autism — often because they did not know much about autism. Instead they collectively called these issues 'blindism.'

To a seasoned eye, the similarities between blindism and autism are striking. They include atypical communication, language and social skills, as well as stereotypies, resistance to change, severe **anxiety** and high **pain** tolerance. And like sighted autistic children, about one in four blind children experiences **regression** at 15 to 30 months of age.

To find out more about this connection, I visited a school for the blind and evaluated 38 of the school's 125 students for autism. I diagnosed autism in 18 of 25 students with congenital blindness but in only 1 of 13 with partial or acquired blindness. A statistical analysis showed that congenital blindness is the main factor responsible for the autism. No other variable, including etiology of blindness, the presence of intellectual disability, overt brain damage or socioeconomic status, accounted for its high prevalence².

As part of this study, I evaluated 12 previous studies of blindness and autism. Each study focused on specific causes of blindness, under the assumption that the participants' autism stemmed from the same cause — say, congenital rubella or optic nerve atrophy. But I found that, taken together, the studies suggest that blindness itself (no matter what its cause) is connected to autism: About half of the collective 859 children who were blind from an early age also have autism. The rate of autism was even higher, ranging from 55 to 74 percent, in children with total congenital blindness.

Special connection:

Some researchers have proposed that the connection between autism and blindness is cognitive. Results published earlier this year suggest that idea is incorrect, however.

This population-based study looked at core autism traits in more than 3,000 adults with intellectual disability, 386 of whom are visually impaired. They found that the visually impaired adults with intellectual disability are more likely to have autism traits than the sighted adults, indicating that the effects of blindness extend beyond intellect. And once again, the prevalence of autism traits is highest among the adults with congenital blindness⁴.

My clinical experience jibes with the notion that intellectual disability is not what binds blindness and autism. There are highly intelligent people who are both blind and autistic. For example, I saw a congenitally blind teenage girl named Brisa who has social and communication difficulties as well as **atypical prosody**, the musical quality of speech. Brisa has excellent reasoning skills and excels academically.

Among the senses, vision also may have a special connection with autism. In 1991, we found that only 46 of 1,150 hearing-impaired children met the criteria for autism. The presence of autism is related more to medical conditions that affect the brain, such as congenital rubella or prematurity, than to the severity of the hearing impairment, suggesting that deafness itself does not contribute to autism³.

Researchers need to nail down the brain mechanisms that account for the autism-blindness overlap. One place to start looking is a brain region called the superior colliculus. This structure receives direct input from the retina. It is involved in not only the **recognition of faces** and **biological movement** but also the integration of sensory input with emotions, basic body functions and motor planning — functions that are often altered in autism⁶.

Another outstanding question is: Why does a small proportion of congenitally blind children develop typically? Early in life, communication is mostly visual in nature, so how do some blind children acquire communication and social skills despite the lack of visual input? Understanding the factors that protect them could provide clues to autism therapies.

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