NEWS

Signs of autism emerge early in babies with related condition

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Infants who have neurofibromatosis type 1, a genetic condition linked to autism, show motor difficulties and communication delays at 10 months of age, according to the first study to thoroughly characterize these babies[1].

The work suggests that motor problems are central to neurofibromatosis type 1, or NF1, and may also be critical to autism. About one in four children with neurofibromatosis **also has autism**.

NF1 is characterized by benign tumors on nerves. Signs of the condition are often evident at birth or soon after, as characteristic spots on the skin. The study is small but suggests that children with the condition also have developmental challenges that start in infancy.

“We saw those delays in both our assessments and in parent reports,” says **Emily Jones**, lecturer at Birkbeck, University of London, who co-led the study. “It’s not something that’s been reported before, but it matches what we see in siblings of children with autism.”

NF1 is typically diagnosed years earlier than autism, making it easier for researchers to track developmental delays in these children.

“This would be a way to study autism when it’s still in formative stages,” says **Anantha Shekhar**, professor of psychiatry at Indiana University in Indianapolis, who was not involved in the work.

NF1 is also easier to study than autism because it is caused by a mutation in a single gene. If the two conditions share features, researchers may be able to mutate or silence the NF1 gene in animals as a window into autism.

“We can potentially learn quite a lot about emergence of symptoms by studying babies at risk for autism because of a monogenic condition,” says **Lauren Weiss**, associate professor of psychiatry at the University of California, San Francisco, who was not involved with the study.

Early intervention:

Jones and her colleagues described findings from six girls and four boys with NF1 when they were 10 months old. The babies are part of an ongoing study of 30 infants with the condition. The researchers assessed the infants’ autism features, cognitive function, language skills and social engagement.

They compared these scores with those from 238 other children, including 166 infants at **high risk of autism** because they have a sibling with the condition. These ‘**baby sibs**’ came from the British Autism Study of Infant Siblings and included 34 children who went on to be diagnosed with autism by age 3.

Infants with NF1 showed delays in language and motor skills, as did the high-risk infants later diagnosed with autism. For instance, the babies would not respond to their name or to simple requests, such as a bid to hand over a toy. Some of the infants could not pull themselves up into a sitting position or grasp objects, as most 10-month-olds can. The work appeared 23 November in *Molecular Autism*.

The findings suggest that language problems in preschoolers with NF1 are present much earlier than experts had thought. They call for a closer look at language development in infants with NF1, so that those deemed at risk of autism can start therapy early. (The researchers don't yet know how many of the children with NF1 have autism.)

"This is an important finding, as it is well established that early intervention is critical to ease autism symptoms," says **Anne Goriely**, senior research fellow at the Weatherall Institute of Molecular Medicine at the University of Oxford in the United Kingdom, who was not involved in the study. "Early recognition and intervention approaches for autism in NF1 families are likely to have a significant impact on the development of these children," she says.

The motor problems are striking, and suggest this is an important domain to study further in animal models, the researchers say. The team plans to add the tracking of eye and head movements to their test battery for infants with NF1 to better characterize motor function in these children.

REFERENCES:

1. Kolesnik A.M. *et al. Mol. Autism* **8**, 62 (2017) **PubMed**