

NEWS

Rare form of autism shows unique pattern of regression

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More than 40 percent of children with Phelan-McDermid syndrome lose skills they once had¹.

Unlike children with autism, in whom regression is reported to typically occur at age 2, children with

this related condition begin to regress at age 6, on average. About 20 percent of children with autism **lose social and language skills**, according to a 2016 study.

The study is the first to examine the prevalence of regression in individuals with Phelan-McDermid syndrome. The condition was first described in 2001 and is caused by a deletion on chromosome 22.

About 84 percent of people with Phelan-McDermid syndrome have autism². The syndrome can also include developmental delay and unusual physical features.

“[Phelan-McDermid syndrome] is its own disorder with its own type of regression,” says lead researcher **Joachim Hallmayer**, professor of psychiatry and behavioral sciences at Stanford University in California. The study appeared 15 March in the *Journal of Psychiatric Research*.

Hallmayer and his team reviewed medical records for 50 people, ages 4 to 48 years, with Phelan-McDermid syndrome. They also asked the participants’ parents whether their children had lost any acquired skills, including motor, language, social and self-help skills, such as feeding themselves or using the toilet.

Of the 50 participants, 18 had clearly regressed, 24 had not, and researchers couldn’t make a determination for the other 8. Boys were nearly twice as likely as girls to experience it.

Controversial concept:

Of the 18 children who experienced regression, 12 lost skills after age 2.5 years, although the timing ranged from as early as 18 months to as late as 18 years. The researchers were not able to examine whether the severity of regression varied with the age of onset, however.

Clinical experience suggests that regression takes on different forms in children than it does in adolescents, says **Catalina Betancur**, director of research at the French National Institute of Health and Medical Research in Paris, who was not involved in the study. “It’s not as severe or pervasive in children as what happens in adolescents and adults,” she says.

Seven of the participants who lost skills eventually regained them, raising questions about what ‘regression’ means. “The idea of regression is still controversial, and we need to follow these children over time,” Hallmayer says.

Regression in Phelan-McDermid syndrome primarily affects motor and self-help skills, the researchers found. Children with autism alone tend to lose language and social skills.

Roughly 63 percent of the individuals in the study have a history of seizures, but Hallmayer and his team found no evidence that regression is associated with seizures as other studies have³.

The researchers point out that the results of the study are not definitive, however. “It was done retrospectively by asking the parents, ‘How was your child functioning?’” Hallmayer says.

Still, the findings provide useful background for clinicians who treat people with Phelan-McDermid syndrome.

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