EARLI
Early Autism Risk Longitudinal Investigation

Fact Sheet

Q: *What is the chance that the younger brother or sister of a child with Autism Spectrum Disorder (ASD) will also have ASD*?

A: It is very difficult to accurately estimate the sibling recurrence risk of ASD. Taking into account all studies to date, the closest estimate lies somewhere between 10% and 25%.

Q: What do we know from past studies?

A: Autism Spectrum Disorder (ASD) is often inherited.

The scientific evidence from studies conducted over recent decades supports the idea that ASD is often inherited. ASD is a term that is used to reflect the range of related disorders characterized by deficits in social interaction, deficits in communication, and repetitive behavior. These include autism, Asperger's Syndrome and PDD-NOS (pervasive developmental disorder not otherwise specified). Studies of twins have shown it is more likely that both twins will be affected when they are genetically identical (monozygous) than when they are not identical (dizygous or 'fraternal'). Other studies that used interviews of family members to track autism back several generations also suggested that autism is heritable, or can be passed on through genes. However, these older studies tended to focus on autistic disorder, as opposed to ASD, and were conducted in an era when the diagnostic criteria for autism differed from those accepted today.

Today, we also know that the genetic component of ASD is quite complicated. Some cases of ASD may be caused by genes passed down across several generations. Up to 10% of ASD cases occur in children who also have another condition of known genetic origin (e.g., Fragile X, tuberous sclerosis). In other instances, ASD may be caused by completely new genetic mutations occurring in the sperm or egg of the parent. It is also possible that other cases of autism might not even have a genetic component at all. It is increasingly clear, however, that several factors are likely acting together to cause the condition.

Q: What do we know specifically about the recurrence of autism in siblings?

A: A younger sister or brother of a child with ASD may also have ASD, but we do not yet know the precise rate of sibling recurrence.

Recurrence is the frequency of ASD in siblings born after the first diagnosed child. Studies of sibling recurrence done in the last twenty years have generally reported that between 2% and 8% of younger siblings of children with ASD were eventually diagnosed with ASD. However, these older studies may not be completely accurate.

Like the twin studies, the majority of these older recurrence risk studies involved only children with autistic disorder diagnoses, and then looked only for autistic disorder in the younger siblings. These studies also tended to include fairly small numbers of families and did not always examine whether the risk of autism in the baby was affected by different characteristics of the older sibling. Estimates from these studies might also be somewhat inaccurate because they did not account for the fact that having a child with autism can change families' decisions about having more children.

More recently, a number of research teams have started to closely follow siblings of children with autism from very early ages. These babies are being closely monitored for the early signs, and later for definite diagnoses of ASD, using today's accepted diagnostic standards. Preliminary results from just two of these efforts have been published and present different findings than the earlier studies.

The first study reported on 107 younger siblings of children with ASD, finding that 31 (28%) met criteria for ASD at 36 months of age. The second reported

on 155 siblings of children with ASD and found that 35 (23%) met criteria for ASD at 36 months.

It is possible that the findings from these more recent studies may over-estimate recurrence because they enrolled siblings after they were born. This suggests that parents who were worried about early behaviors that they already observed in their babies may have been most likely to participate. Therefore, it is still very difficult to accurately estimate the sibling recurrence risk of ASD. The true estimate probably lies between the older and newer estimates – somewhere between 10% and 25%.

Q. How will EARLI help us to better understand sibling recurrence?

A. The main objective of the EARLI Study is to understand autism's complex causes and to identify potential risk factors.

EARLI will also provide new information about the sibling recurrence of ASD. We hope to eventually have data on the ASD status of at least 1,000 younger siblings of children with ASD, making the EARLI Study larger than other studies. In addition, because families are enrolled when the mother is pregnant, there is less opportunity for EARLI's findings on sibling recurrence to be influenced by 'selection bias' — when parents who are already worried about their baby's behavior are more likely to participate in the study.

Because EARLI will be recruiting and following families for several years, it is likely that new findings from other research on sibling recurrence will emerge while the EARLI Study is still in progress. The EARLI team will be monitoring the scientific literature for these findings and will communicate information to our participants through the study website and through study newsletters. In addition, this factsheet will be updated when additional evidence accumulates.

EARLI
www.EARLIStudy.org

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