Autism: Findings from Postmortem Brain Tissue Research

Autism spectrum disorder is a neurodevelopmental condition defined by challenges in social interactions and restricted or repetitive behaviors and interests.

Autism is linked to changes in the structure and function of the brain. Research using donated postmortem brain tissue helps researchers identify cellular, genetic, and molecular changes in the brains of autistic people.

Cellular Research: Key Findings

Different types of cells in the brain have been implicated
- Neurons – cells of the nervous system that process information\(^1-5\)
- Glial cells – cells that support the function of neurons and support the brain’s response to injury and inflammation\(^6-8\)

Neuron structure, number, connectivity, and neurotransmission
- Changes in the number and size of neurons in several regions of the brain\(^1-5,9\)
- Changes in the number of connections on individual neurons\(^10\)
- Changes in neurotransmitters, which are brain chemicals that excite or inhibit communication between neurons\(^11-13\)
- Changes in brain chemicals, such as oxytocin that regulate social behavior, sleep, appetite, and emotion\(^14-15\)

These findings suggest that autism affects a variety of cell types and circuits, that control brain communication and the immune response in the brain.

Genetic and Molecular Research: Key Findings

DNA sequence and activity
- Changes in the DNA of brain cells that occur during fetal brain development. These changes, which are called "somatic mutations," can affect parts of the DNA that control the activity of genes and may increase a person's chance of having autism\(^16\)
- Changes in DNA sequence that results in altered proteins\(^17\)
- Decreased activity of genes that control neural communication\(^17\)
- Increased activity of genes that regulate immune functions\(^17\)

Gene regulation
- Changes in epigenetic processes that affect how genes are turned on and off\(^18\)

These findings suggest that in autism, changes in gene activity can affect different aspects of brain functioning, such as neural communication and immune responses. Some changes may be due to genetic mutations, whereas others, called epigenetic changes, may be caused by interactions between the genes and the environment.
Future research

Many questions remain unanswered. Goals of future research include:

- Understanding how cellular, genetic, and molecular changes relate to the traits of autism and associated neurodevelopmental conditions.
- Identifying which biological changes are specific to autism and which changes are shared with psychiatric conditions, and other comorbidities.
- Using the knowledge from brain tissue research to develop targeted treatments to decrease the challenges of autism.

The overall goal of brain tissue research is to improve the quality of life of autistic individuals.

This research is made possible by the gift of brain donation to Autism BrainNet and other brain tissue repositories. To learn more about autism research using postmortem brain tissue visit https://www.autismbrainnet.org/category/science-articles/.

REFERENCES


What is Autism BrainNet?

Autism BrainNet is a program of the Simons Foundation Autism Research Initiative (SFARI) that promotes innovative, high-quality research on postmortem brain tissue with the goal of improving the understanding of the biological causes of autism spectrum disorder and related neurodevelopmental conditions.

Contact Us

To learn more: Visit autismbrainnet.org or call toll-free at 877-333-0999. We are available 24/7.

For general inquiries about Autism BrainNet: Email us at info@autismbrainnet.org

For questions about the tissue application process: Email us at requests@autismbrainnet.org

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