

Interview with Dr. Martha Herbert— Autism: a brain disorder or a disorder that affects the brain?

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Abstract

Autism research priorities have been shaped by underlying models. The model of autism as a genetically determined hard-wired brain disorder, dominant in recent years, has led to a search for “brain genes” and brain alterations. But this model has produced limited results, has rested on an over-interpretation of evidence for heritability, and has also failed to encompass multiple features of autism outside the behavioral definition, including systemic physiological changes (especially, but not restricted to, gastrointestinal and immune) and the increasing numbers of cases. A more inclusive model would construe autism as a disorder that affects the brain, and that is the outcome of complex interactions among factors related to genetic vulnerability, environmental triggers or causes, and epigenetic changes. This model can incorporate many recent findings, and it opens the field on several levels: to broader genetic investigations (including, for example, systemically expressed genes that could impact the brain secondarily), and to study of vulnerabilities beyond genetics at multiple physiological levels. Since the behaviors that define autism appear to be produced by brains affected by a variety of biological alterations, this more inclusive model is also better oriented to encompassing autism's heterogeneity. It allows us to investigate what systems and network-level commonalities there might be among brain and body changes whose specific biological details may differ. Of paramount practical importance is that some features of systemic involvement may be modifiable. Thus, we may therefore more aggressively search for such features as potential treatment targets that may reduce suffering and improve options for affected individuals. By improving metabolic status, parameters modulating brain function (e.g., synaptic thresholds, connectivity, energy metabolism) may be affected in a favorable way. This may account for some of the growing number of anecdotal reports of recovery from autism after integrative biomedical and behavioral treatment. Moving from a “genes-brain-behavior” to a “pathogenesis (genes, environment, epigenetics)-mechanism (molecular, cellular, tissue, processing)-phenotype (behavior, sensory-perceptual, cognition, medical)” model, which not only spells out the levels of the biological hierarchy, but also looks at all these levels developmentally, is a challenge to compartmentalized science, but this is what we need if we are to translationally connect research and successful treatment.

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