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Sommario/riassunto	This Research Topic will review and summarize the pathogenesis of Autism Spectrum Disorder (ASD) that underpin the translation of genetic vulnerability to clinically significant symptoms. Available research data in ASD suggests that it is a “neural connectivity disorder” and that the deficits in social cognition and related neurocognitive functions result from reduced synchronization between key brain regions known as the “social brain”. These interconnected neural systems can be understood through the relationship between functionally relevant anatomic areas and neurochemical pathways, the programming of which are genetically modulated during neurodevelopment and mediated through a range of neuropeptides and interacting neurotransmitter systems. Elucidating the underlying molecular mechanisms can provide an invaluable window for understanding the neural wiring that regulates higher brain functions and consequent clinical phenotypes. ASD is a heterogeneous condition

and clinical heterogeneity is linked to genetic heterogeneity. Phenotypic variability within ASD and the phenotypic overlap between ASD and other neurodevelopmental disorders such as Tourette Syndrome, ADHD, Schizophrenia, language disorder and intellectual disability could be associated with the fact that the genes converge on a common neurodevelopmental pathway involved in synapse development/maintenance and circuitry formation through effects on neurogenesis, axon guidance in dendritic projections or neuronal migration. Thus defects in synaptic development can result in abnormal development across disorders and broad domains but yet carry distinct neurocognitive and behavioral profiles. The penetrance of the different co-morbidities may in turn be related to the dose effects of gene abnormality or the timing of events when different neuronal regions and circuitry are being formed, as may be the influence of gender, intrauterine and perinatal events, epigenetics and other environmental modulators. In keeping with the multi modal and diverse origins of neurodevelopmental disorders, this review will explore the genetic underpinnings and environmental modulation in the aetiology; neural substrates, biomarkers and endophenotypes that underlie clinical characteristics of ASD; as well as neurochemical pathways and pathophysiological mechanisms that pave the way for therapeutic interventions. Furthermore, since genetically mediated deficits and consequent functional impairments involve activity-dependent synapse development that depends on postnatal learning and experience, early intervention can prevent or reduce the risk of these deficits cascading into a trajectory toward full expression of the disorder by exploiting the neuronal maturation and brain plasticity. In addition to reviewing the current state of evidence in the literature, there will be a significant focus on ongoing original work as well as hypotheses and directions for future research.

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