

What Underlies Autism Spectrum Disorder?

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Autism spectrum disorder (ASD) is an early onset, mostly life-long pathological condition characterized by significant deficits in social communication skills and in the range of interests/activities.¹⁻³ A multifactorial etiology with the prevailing weight of genetic factors is hypothesized, but the high increase in the prevalence of this condition in recent decades⁴ leads to speculate a significant role also of environmental factors including air pollutants.^{5,6} A question arises spontaneously in the professionals who deal with these individuals: regardless of the etiology of ASD, which unfortunately is still unknown in most cases today,⁵ what is the basis of the core symptoms that characterize autism? Is there a single pathogenetic mechanism, common to all cases, or is there more than one? Answering these questions would have very important implications both in terms of prevention in cases so-called at high risk of developing autism (see for example the siblings of children with ASD)⁷ and in terms of enabling intervention for individuals who have already received the ASD diagnosis. There are basically 3 main hypotheses that have been elaborated so far to explain the core symptoms of autism: each of them helps to interpret some of the symptoms that characterize this condition, but not all. Here the reference is made to the possible dysfunction of the theory of mind,^{8,9} of the executive functions,^{10,11} and of the central coherence.^{12,13}

Theory of mind (ToM) relates to the skills needed to interpret others' behaviors based on their mental states, including feelings and beliefs, and to identify the intentions and emotions of others, as well as self-awareness. ToM skills' impairments can cause social communication deficits in ASD children due to the lack of perception that behaviors are driven by mental states.^{8,9} The so-called mirror neurons are a functional set of cortical neurons activated both by performing an action and observing the action performed by others. Mirror neurons are mainly located in the inferior frontal, premotor, supplementary motor, primary somatosensory, and inferior parietal cortex. They are hypothetically related to social skills, including ToM, and their dysfunction in ASD could impair the understanding of others' actions and intentions. Therefore, they could represent, at least in part, the neurobiological substrate of the ToM.⁹

Executive functions are a set of cognitive processes very important for the ability to adapt to new environmental stimuli, particularly if a new behavior is necessary. They include inter alia, attention, working memory, planning, inhibitory control, and cognitive flexibility and are particularly related to prefrontal cortical areas. Executive functions, therefore, appear to be widely involved in social cognition skills and adaptive behavior, and their impairments may explain some autism core features.^{10,11}

According to the weak central coherence (WCC) theory, ASD individuals use a detail-oriented information processing style that is effective in tasks involving visual search, but it is much less effective when coherence is helpful, as in social cognition tasks.^{12,13} In ASD individuals engaged in locally directed visual tasks, brain activity has been related to the inferior frontal gyrus and to parietal regions. In addition, excessive reliance on posterior, basic visual processing cortical regions has been suggested in these individuals, as well as reduced functional connectivity between anterior and posterior regions. This processing bias, which leads to excessive attention to detail, could impair social interactions, which require the correct integration of different elements such as mimicry, voice, gestures, and environmental context.¹³

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It should also be emphasized that these 3 hypotheses do not exclude each other and each of them seems to explain only a part of the complex autistic symptomatology.

But there may be also other interpretations of the behavior of an individual with ASD. Here we refer to possible sensory abnormalities and particularly an impairment of multisensory (or multimodal) integration, which is probably due to altered brain long-range connectivity. According to this hypothesis, ASD individuals have a lesser ability to integrate sensory information from different modalities (visual, auditory, tactile, etc.).¹⁴ In this regard, a highly multimodal experience is represented by joining a conversation or in general social interaction with another person. In this situation, we have to take care of several visual, auditory, and sometimes other stimuli at the same time,¹⁴ processing them correctly and quickly in order to adequately interpret the messages (both verbal and non-verbal) of other persons and to give them an answer in a timely and sufficiently understandable manner. It is easy to imagine how this situation can put an individual with ASD in serious difficulty, especially when he/she is younger, even if he/she has adequate (or even above the norm) intellectual functioning, also causing him/her a considerable amount of discomfort and stress and consequently a low motivation or even an active refusal to engage in social interaction.¹⁴

Understanding the mechanisms underlying the behavior of an individual with ASD is fundamental also and above all in the perspective of carrying out enabling interventions aimed as much as possible at facing his/her difficulties and at meeting his/her needs in everyday life. And, indeed, there are data in the literature in favor of the efficacy of enabling interventions aimed specifically at acting on the mechanisms that are hypothesized to be involved in the development of autistic symptoms (see above).^{11,15,16} But could there be a common element underlying the various theories reported above? An impairment of sensory filtering, for which involvement of the brainstem and/or atypical subcortico-cortical connectivity is assumed,^{17,18} could be hypothetically the core mechanism underlying autistic symptoms because it does not allow the ASD individual to identify and value the significant elements of what surrounds him/her leaving the less important elements in the background. Therefore, altered sensory filtering could explain at the same time the difficulties in interpreting others' behavior based on their mental states (see ToM),^{8,9} in adapting to new environmental stimuli (see the theory of altered executive functions),^{10,11} in processing visual information (see the WCC theory),^{12,13} and in integrating sensory information from different modalities.¹⁴

These reflections suggest a possible key to interpret the behaviors of individuals with ASD in daily life and at the same time offer insights that could prove useful in terms of enabling intervention.

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