An overview of theories that address the evolution of autism.

Autism as the result of an extreme male brain

The extreme male brain theory of autism postulates that affected individuals are extremely focused on systemizing as opposed to empathizing.

Men, appear to have a more systemizing brain than women.

They are more interested in and better at analyzing variables in a system, and at deriving the rules that govern the behavior of a system.

Women, on the other hand, seem to have a more empathizing brain, they are better at inferring mental states in other people, and to respond appropriately to these mental states.

Empirical support for the extreme male brain theory of autism comes from several sources.

First, more males are affected by autism than females.

Second, high-functioning affected individuals (i.e., with an average or above average IQ) tend to outperform unaffected people with similar IQs on systemizing tasks.

Third, the behavioral differences between people with and without autism are mediated by differences at the anatomical level of the brain.

Fourth, prenatal exposure to testosterone (an androgen) is positively related to the development of autistic traits.

So having an extreme male brain, a condition which we strongly associate with autism, may have had practical advantages given demands of ancestral times for developing tools and weapons, in hunting, tracking, and trading.

Autism as the result of an extreme imprinted brain

A second theory on the evolution of autism is the imbalanced genomic imprinting theory.

Genomic imprinting refers to the expression of genes from only one of the two parental chromosomes.

We inherit two copies of every allele, a maternal and a paternal copy. In most cases both copies are functional, but in some exceptional cases one of the copies is turned off and thus not functional.

This may be the consequence of imprinting: maternal imprinting ensures that only the maternal copy is expressed, and paternal imprinting ensures that only the paternal copy is expressed.

The evolutionary function of imprinted genes is unknown. It has been suggested that genomic imprinting originates in a conflict between the sexes about the amount of investment of the mother in the child.

Paternally expressed imprinted genes tend to promote fetal growth, whereas maternally expressed imprinted genes tend to suppress fetal growth.

From the father's point of view, it is beneficial that the mother invests as much as possible in the child. From the mother's point of view, it is important to preserve her resources. This implies that it is beneficial for her to invest in the child only as much as is necessary.

It has been shown that imprinted genes are highly expressive in the central nervous system, and that they are involved in neurodevelopment.

Imprinted genes are often implicated in disorders, because a single change can dysregulate their function. Genomic imprinting has been linked to several disorders, including autism.

Crespi and Badcock suggested that children with autism impose additional demands compared to normal children, especially on mothers, who tend to be the primary caregiver. Normal children who display such behavior also impose additional demands on the mother, which is beneficial from the point of view of the father, because the mother will spend more of her time and resources on the child.

However, in the case of autism, the behavior of the child assumes pathological proportions which no longer benefit either the mother or the father.

Autism as the result of a reptile brain

A different perspective on the evolution of autism is provided by the Polyvagal theory. The theory postulates that through three stages of phylogeny, mammals, especially primates, including humans, have evolved a functional neural organization that regulates emotions and social behavior.

It is proposed that people with autism minimize the expression of the mammalian response, i.e., social communication.

While normally primates and humans have a well-developed ability to shift adaptively between mobilization and social engagement behaviors, individuals with autism lack this ability.

The resulting behavioral features lead to adaptive benefits in focusing on objects, while minimizing the potentially dangerous interactions with people.

The combination of a nervous system that favors defensive behaviors, and the inability to use social communication with people, places the autistic individual outside the realm of normal social behavior.

Thus, due to the inability to engage the myelinated vagus to calm and dampen the defensive system (through social interactions), the nervous system of the autistic individual is in a constant state of hypervigilance or shutdown.

These are generally adaptive responses in reptiles, but are severely maladaptive in mammals. It does not explain the genetic background and the heritability of autism, so the reptile brain theory needs some extension to give a full explanation of the evolution of autism.

Autism as the result of epistatic interactions between the effects of genes

Epicastic = an interaction in which one gene suppresses the expression of another

It is well-established that autism is caused by many interacting genes. As nearly 30 genes have been associated with autism, autism is clearly no Mendelian (single gene) disorder.

An evolutionary theory of autism should take into account the developmental effects of both its polygenic nature and of interactions among the genes (i.e., epistatic interactions).

Assume that there are 30 genes involved in the development of autism (this number is likely to be larger), and that this same set of genes is involved in the development of intelligence.

Given the evidence that intelligence is positively correlated with potential reproductive success, the 30 genes that are involved in autism can potentially spread in the population, thanks to the link with intelligence. In most people, interactions between these 30 genes result in an individual with normal or high intelligence, without autism.

However, some unlucky interactions, especially in combination with negative spontaneous mutations, lead to the development of autism, low intelligence, or other pathologies.

On certain intelligence tests, individuals with autism show equal or better performance levels compared to normal individuals. There is also evidence for the relation between autism and exceptional abilities, with some famous examples of autistic savants.

The co-occurrence of savant syndrome and autism is an example of the effect of epistatic interactions between genes, in which potentially beneficial effects of genes are nullified by the negative effect of autism.

The proposal that the combination of high heritability and low fertility in autism can be explained by the effects of epistatic interactions between genes that are involved in both intelligence and autism.

The Theory of Mind Hypothesis of autism

Theory of mind has been an area of significant interest in recent autism research as it is such a prominent deficit in individuals diagnosed with autism. Theory of mind refers to the ability to infer the full range of mental states (beliefs, emotions, desires, intentions, imagination, etc.) that cause action.

Possessing a theory of mind is to be able to effectively reflect on the contents of one's own and others minds. It also allows us to predict and anticipate behaviors in others and respond accordingly and appropriately. Difficulty in understanding other minds and interpreting behavior is a core cognitive feature of individuals diagnosed with autism.

In essence, this theory states that individuals with autism fail to "impute mental states to themselves and others" and that this deficit manifests as inability to mentalise, or failure to take into account others' mental states.

The most widely used test of Theory of Mind is the unexpected transfer test of false belief. In the task the participant watches a sequence of events, usually enacted by dolls. The story unfolds so that one doll has a belief about the location of an object that is incongruous with its real location.

The participant then makes a judgement about where the doll will look, and in order to give the correct answer the participant has to infer the mental state of the doll (I think he thinks). 80 percent (16/20) of children with autism failed the unexpected transfer task and concluded that these children had a deficit in their theory of mind.

Happé, stated that it was problematical for the Theory of Mind Hypothesis that 20 percent of autistic individuals actually passed tests of false belief, and so the deficit seemed not to be universal. So the Theory of Mind Hypothesis may explain some of the cognitive impairments seen in autism, but that it does not fully explain all facets of the disorder.

In the face of the issue of universality, they used the more difficult second-order false belief task (I think he thinks she thinks), and none of the children with autism passed the test. Baron-Cohen concluded that although some individuals with autism may be capable of passing a first-order theory of mind task, they could not pass a second-order task and therefore did not have a fully representational theory of mind.

The theory of mind account of autism has been remarkably successful in making specific predictions about the impairments in socialization, imagination and communication shown by people with autism.

It cannot explain either the non-triad features of autism, or earlier experimental findings of abnormal assets and deficits on non-social tasks.

These unexplained aspects of autism, and the existence of autistic individuals who consistently pass false belief tasks, suggest that it may be necessary to postulate an additional cognitive abnormality.

Weak Central Coherence theory

Weak Central Coherence Theory is a domain general process, and one of its key strengths is that it explains some of the non-social, as well as the social features of autism, such as the attention to acute detail that ranges from pedantry to obsession.

The essence of the theory is that typically developing individuals process information by extracting overall meaning or gist. Frith and Happé suggest autism is characterized by weak or absent drive for global coherence.

That is, individuals with autism process things in a detail-focused or piecemeal way—processing the constituent parts, rather than the global whole.

The broken mirror theory

According to this view, autism-related disorders might be caused by a hypoactivity of mirror neurons, a neuronal system that is activated when an action is performed by a person, and when the subject observes the same action done by a conspecific.

These neurons are one of the key mechanisms for what concerns social interactions, as it allows an individual to embody in himself the mental states of those who have faced as they were their own.

Autistic patients would not be able to embody in themselves others' mental states (intentions, beliefs, expectations, etc.) and this would be due to a dysfunction related to mirror neurons.

Even if much evidence supports this theory, however, there is also a large number of studies demostrating that the hypoactivity of the mirror system would be found only in certain circumstances and not in other.

It has been shown that in imitation tasks which do not require an explicit imitative behavior, there was a hypoactivity of the autistic mirror system. However, this does not occur when the subject is explicitly asked to imitate an observed movement.

These patients also have difficulties for certain types of actions (e.g., actions without a goal), but show a normal behavior with others (e.g., direct action directed to a goal).

Moreover, the deficit associated with the mirror system does not stop at simple task imitation. Autistic subjects, in fact, prove to have great difficulty in understanding the others' intentions.

Indeed, they seem to have no impairment regarding the goal of the action, or rather the "what" of the action. What they could not understand is the general intention of the model, or rather "why" this action is performed.

Conclusions: Autistic subjects, for those problems of embodiment of the mental states of others, which are possible only thanks to mirror neurons, have many difficulties in reading the emotional states of others.