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Autism spectrum disorder-from genetics to intervention

Abstract

The Autism Spectrum Disorders (ASD) is developmental disorders that affect individuals at different levels - behavioral, linguistic and social–and in different magnitudes (APA, 2013).[1] This is a disorder that has been studied a lot in recent times, due to the exponential increase in cases. However, it continues to leave many in conclusions due to its heterogeneity. In this article we intend to create an overview of this problem, from its causes to the intervention that still obtains better scientific results, the ABA behavioral therapy (Applied Behavior Analysis).

Keywords: autism, genetics of autism, childhood disintegrative disorder, applied behavior analysis, behavior modification, language development

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Daniela Vieira, Ana Paula Couceiro Figueira
Faculty of Psychology and Educational Sciences of the University of Coimbra, Coimbra, Portugal

Correspondence: Daniela Vieira, Faculty of Psychology and Educational Sciences of the University of Coimbra, Coimbra, Portugal, Email danii_vieir@live.com.pt

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Introduction

Currently, there is still no specific biological marker that identifies Autism Spectrum Disorder (ASD) or Autism (as it's commonly identified, and your heterogeneous.[2] Thus, the diagnosis is based on the guidelines described on DSM-5 (Diagnostic and Statistical Manual of Mental Disorders 5th edition) and in ICD-11 (International Classification of Diseases 11th revision).[3]

In 2012, it was estimated that, on average 100/10.000 people worldwide would have autism.[4] In 2014, studies confirmed the affected one's to be 1 in every 59 children, up to eight years of age, validating an exponential growth.[5]

Genetics of autism

Despite not being able to identify a specific cause for ASD, (0,65-0,91) studies based on families with twins confirms its relation to heredity.[6]

There are two points that stand out, "First, it is now appreciated that the new mutations contribute to ASD and often carry large effects. Second, the advent of next-generation sequencing technologies has enabled hypothesis- naive whole-exome surveys of large ASD cohorts to identify genes with the new, ASD- associated damaging mutations".[7] In this study it was noticed that the genetic mutations identified are revealed in the diagnostic criteria for ASD. On the other hand, new generation technologies for brain analysis have made it possible to expand ASD investigations, however, they have not identified any genome that would support the appearance of a new mutation and consequently trigger a new case of ASD.[7]

Despite of Several studies, the concept of ASD still remains a puzzle due to diagnosing difficulty.[3,8]

Brain regions such as the cerebellum, temporal lobe, fusiform gyrus, amygdala, frontal lobes and white matter are the most identified with problems within the neural network of this psychopathology, however, again, without consistency for all cases.

The investigation also suggests higher prevalence the conception of the fetus. The disorder arises from the abnormal development of the neural circuits and from the connections between the brain network systems".[2] Yet, not all cases are identified with these criteria from birth, as we will verify on the next topic.

Childhood disintegrative disorder

Theodor Heller, in 1908, identified a group of children, between 3 and 4 years old, who had a typical development and subsequently had a sudden and severe regression of speech and cognition. This psychopathology became known as Childhood Disintegrative Disorder (CDD) according to ICD-10 and DSM-IV-TR.[9]

In the Diagnostic and Statistical Manual of Mental Disorders (DSM-V), some concepts relating to ASD were changed. One of the most glaring changes included the creation of the term autism spectrum disorder, which included what was previously known as Childhood Disintegrative Disorder. In "cases where there has been a loss of skills, parents or caregivers may report a history of gradual or relatively rapid deterioration in social behavior or language skills." The manual also says that in "this will occur between 12 and 24 months of age. Age, being distinguishable from rare cases of developmental regression that occur after at least 2 years of normal development."[1]

However, "literature reviews had different conclusion about the validity of CDD as a separate entity to autism spectrum disorder (ASD)".[9]

A higher prevalence of epilepsy, mutism and intellectual disability stands out high in CDD compared to ASD. The CDD is also associated to a broader set of impairments than the ASD such as adaptive skills and emotional and behavioral regulation. A study carried out by Mehra et al.,[9] recommends that more than 90% of participants with childhood disintegrative disorder (CDD) have intellectual, language, and social impairments; CDD and autism spectrum disorder (ASD) share features of core and extra- diagnostic symptoms; CDD has more severe impairment than ASD (e.g. lower IQ, more epilepsy).; Differences in CDD include faster regression, more mental health symptoms, and more global developmental deficit.; and gene expression profiles may differ between CDD and ASD.[9]

Another point to note is that most ASD regressions involve a deficit in social communication development prior to the regression, while in CDD, the first two years of life seems to be crucial and causes the onset of the disorder. Last but not least, "regression in CDD has been described as rapid and dramatic, distinguishing it from the more gradual regression process in ASD." If we take into account a less severe level of ASD, we could say that CDD is more severe than ASD. However, some researchers argue "that CDD is part of a continuum

within ASD. Some reviews suggest that, after regression, CDD is 'essentially identical' to ASD: phenotypic similarities include both core social-communication impairments and comorbid intellectual disability and epilepsy. Further, regression, a salient feature of CDD, has also been described in ASD, including in prospective longitudinal studies. A meta-analysis of 29 035 participants with ASD found a 32% prevalence of regression of developmental and/or adaptive skills".[9]

Applied behavior analysis (ABA)

"The purpose of the intervention (...) is to create alternatives and promote adjustments, starting from the real model of the subject's representations, identifying and respecting his/her characteristics, using his/her abilities to the maximum and elaborating adaptive strategies to better deal with his/her limitations".[10] When we refer to the real model, it means being the closest to the subject's day-to-day reality. For example, let's imagine a child who doesn't speak and is always running around, in one family the priority may be to promote communication either through speech or an alternative communication system, however, for another family the priority may be to remain quiet and in that case In this case, the intervention will focus on increasing waiting periods, for example, sitting.[10]

However, there are several therapies that aim to empower individuals with ASD, in the most varied areas. Among the most diverse therapies, the Behavior Modification Model stands out, namely, the behavioral intervention ABA - Applied Behavior Analysis, scientifically proven and to date with better evolutionary results.[11]

This model emerged in the early 1960s. However, it was with Lovaas (1981) that the first behavioral intervention for autism emerged. This intervention tried to adjust the child's behaviors so that he or she would function correctly at school and at home.[12]

The program was 40 hours per week, for two or more years. These programs are personalized for each subject according to their difficulties and abilities. However, the goals are always to increase undesirable behaviors and decrease maladaptive behaviors. The level of demand increased according to the successes obtained (Marques, 2000). Yet, this model is criticized for the difficulty of generalizing what is learned to other contexts and situations. Parents were asked to "adopt a didactic posture in the transmission of acquired knowledge in order to facilitate the generalization of skills trained in the classroom".[12]

ABA as a science was established in the early second half of the 20th century as an approach for the assessment and selection of human behavior change, based on the principles of B. F. Skinner's operant conditioning (Kearney, 2009). "Applied behavior analysis is an approach to modify social behaviors based on scientificprinciples".[13]

Lovaas provided intensive behavioral intervention for children with autism, using the principles and strategies developed by Skinner and his colleagues in the area of the application of behavior analysis.[13]

The ABC model is "taken" from Skinner's theory in which: A stands for antecedent, the stimulus that occurs before the behavior, that is, what initiates the behavior; B from behavior, everything that the individual does after the antecedent, that is, the individual's response; C from consequence, what occurs after the behavior, after the individual's response. The antecedents are detected by the senses and can be neutral or indicate reinforcement or punishment. The behavior is what is intended to be changed either in order to increase or decrease it. The consequence is immediate to the behavior, if it is regular; it has an effect on the frequency of occurrence.[1]

ABA and the behavior modification

In order to proceed with behavior modification, it is necessary to understand which variables determine the behavior, because in the case of inappropriate behavior (aggression, self- injury, avoidance, stereotyping, tantrums), these variables can maintain it. Among the variables that maintain the behavior the ones that stand out are: 1) attention; the subject receives attention(a wake-up call, a look, etc.) after performing an inappropriate behavior; 2) avoidance/escape: the subject may end or avoid a situation if he/she has inappropriate behavior (e.g. he/she cries a lot so the mother stops insisting that he/she picks up the paper); 3) stimulation: the individual may self-stimulate if he or she is misbehaving; 4) attaining objects: he/she can get back what was taken away from them (e.g. the toy) if he/she behaves appropriately; 5) multi determinate, inappropriate behaviors that have as a consequence several of the aforementioned (e.g., attention and object gain).[14]

First of all, one must understand the variables that maintain a certain behavior, and one must understand the meaning of that behavior (functional analysis).Finally, teach other more appropriate forms of communication.[14]

There are four key steps to ABA intervention: 1) initial assessment; 2) defining goals to be achieved; 3) implementing stipulated programs; 4) evaluating progress.[14]

In ABA behavioral therapy, the first step of intervention is assessment, which is carried out through an interview. This interview is carried out with the subject's reference people (parents, teachers, and other professionals). The goal of these interviews is to understand the individual's learning history, needs and priorities, as well as their expectations of therapy (and other professionals). The goal of these interviews is to understand the individual's learning history, needs and priorities, as well as their expectations of therapy.[15] This first part also includes the application of tests (VB- MAPP, ABLLS-R, ECA, etc.). The main goal is to identify the "behavioral repertoire" in each area of development.[15]

Throughout this process, observation occurs, and should be descriptive and direct, and takes place not only in the clinical setting, but also in the subject's natural environment (e.g., at school). The goal is to identify target behaviors and the possible variables that affect it.[15]

After understanding what the child is capable of doing, we should draw up an intervention plan. The goals to be achieved can be basic, intermediate, or advanced. Basic goals are, for example, to increase eye contact time, to increase the amount of time the child remains seated, to do object recognition, among others. The intermediate ones concern the achievement of daily activities (e.g., brushing teeth, brushing hair, etc.), these activities should be divided into small steps, explained to the child and reproduced/played (learning by modeling), the child only moves on to the next step after successfully completing the previous one. Finally, advanced goals relate to a more intellectual scope (e.g., perception of emotions, calculation, etc.).[15] However, it is not only important to teach new behaviors, but also to decrease those that are maladaptive (e.g., aggression, self-aggression, destruction of the environment, tantrums, etc.).

ABA programs start with an instruction (e.g., point to the apples), if necessary the prompt is used (one should start with the most effective to the least intrusive possible). Planning should be made for a gradual removal of the prompt (prompt fading) to promote response independence.[13]

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After the child has performed the initial instruction, he or she is given a reinforcer (e.g., a sweet or a toy) that should be the most appropriate for the task. There should be a reinforcement scheme to progressively decrease the frequency of reinforcement to fit the child's natural environment.[13] Throughout therapy one must determine what one wants to assess: percentage, frequency, duration, and magnitude of the behavior.[13]

In ABA therapy the assessment is not only exhausted in the initial phase, this assessment should be continuous and systematic, thus it occurs in all sessions (Piñero-Ortiz & Toro-Herrera, 2012).

The evaluation record is made session by session so it can be possible to understand the small evolutions that are being made and, in this way, it is possible to increase the degree of difficulty. On the other hand, if the program is not working, the strategy can be changed (Piñero-Ortiz & Toro- Herrera, 2012).

According to, Marques 2000, this intervention has brought great benefits for children in early stages of intervention and under the age of five.

Given the good results obtained with ABA therapy, the hypothesis of combining it with technology is now being studied, and a technological rehabilitation system following ABA guidelines has already been created.[16]

Language development

Abnormal language development is not part of the diagnostic criteria for ASD, however, early language delays remain one of the warning signs[17] and "stereotyped and repetitive use of language or idiosyncratic language," are considered criteria according to DSM-5for diagnosis.[18]

In most children with ASD, there is a delay in language acquisition. Comprehension and pragmatics are invariably affected. "Comprehension and pragmatics are invariably affected. Lower level mixed receptive/expressive disorders involve phonological and syntactical processing, whereas higher level processing disorders involve semantics and formulation of discourse. In some children, lower-level disorders may be so severe as to preclude speech, where as in others phonology maybe deficient in spontaneous production but not in repetition. Abnormal features of autistic language include aberrant prosody, immediate and delay edecholalia (scripts), and perseveration"[19]

According to several studies, communication impairment is related to impaired social interaction, even when language is present (for example, subjects with ASD exhibit one-sided,non -reciprocal conversation). The language disorder itself, when present, is not limited to difficulty in acquiring spoken language, the ability to acquire sign language is usually equally affected.[20]

The most debated hypothesis about the impairment of language acquisition is that it is caused by "the social and emotional malfunctions which prevent people with autism from developing a 'theory of mind'. This explanation rests on the argument that early stages in the development of a theory of mind are necessary for the ability to use symbols, such as words or manual signs. This hypothesis, however, has difficulty in explaining why people with AS do acquire language, even in the absence of the usual early-developing theory of mind skills".[20]

Another hypothesis about this impairment states that there is a "deficit in the ability to process transient, sequential stimuli, i.e., stimuli with a temporal dimension, such as speech or manual signing".[20] The latter hypothesis is not as widely discussed, however, the mechanisms behind both hypotheses are related to psychological causes.

Conclusion

The last decade has seen an exponential increase in cases of ASD, as well as great progress in its research.[4] To this day, no cause for autism has been found. However, there are several theories that seek an explanation about its origin.[10] Lately, genetic analysis has been used and the hypothesis of the mutation of some genes has been studied, however, without coherent results. Although the diagnosis of ASD includes what was previously referred to as CDD, studies suggest that there are some differences.[9] Because there is no cause for autism spectrum disorders, there is also no cure.[10] However, there are several studies that indicate ABA behavioral therapy as the one with the best results.[16] Language development is implicated in most cases of ASD, however, it is not a diagnostic criterion and the theories underlying its deficits are related to psychological causes.

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Conflicts of interest

Authors declare that there is no conflict of interest.

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