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Genetic counseling and autism: new perspectives for diagnosis and intervention

Aconselhamento genético e autismo: novas perspectivas para diagnóstico e intervenção

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ABSTRACT

Introduction: Autism Spectrum Disorder is a neurodevelopmental disorder of multifactorial origin, affecting social, behavioral, and communication skills from early childhood. With a global prevalence of 1.25%, genetic counseling is crucial to understand the genetic factors involved and estimate the risk of new cases within the family.

Objective: To analyze the importance of genetic counseling in the context of autism, focusing on genetic factors and the recurrence risk of the disorder.

Method: A systematic literature review was conducted in PubMed, SciELO, BVS, Lilacs, and Google Scholar databases, in Portuguese, English, and Spanish. The search included descriptors as "autism", "genetic counseling", "genetics", and "recurrence risk", resulting in the selection of 26 relevant articles.

Result: Most cases of Autism Spectrum Disorder are genetically determined, making genetic counseling essential for assessing recurrence risk and the genetic and environmental factors involved. The recurrence risk varies according to the degree of kinship, being higher among close relatives. Heritability of the disorder is more pronounced in monozygotic twins, and an increased risk is also observed among siblings and in families with other cases of Autism Spectrum Disorder or Attention Deficit Hyperactivity Disorder. The former is more prevalent in boys. Maternal effects during the prenatal period do not show significant influence.

Conclusion: Genetic counseling is essential for clarifying the disorder and is important in both public and private sectors to inform families about recurrence risks and the genetic factors involved, given its high prevalence in the general population.

KEYWORDS: Autism spectrum disorder. Genetic counseling. Genetics.

Central Message

Autism Spectrum Disorder (ASD) is one of the neurodevelopmental disorders with the fastest growing incidence in the world. An estimated 1 in 36 American children have ASD. This incidence increases when there is more than one case in the family. This issue generates intense suffering, especially for parents, who have increasingly sought genetic counseling services to clarify their doubts and know the probability of family recurrence. This review addresses the prevalence, risk of recurrence, and the importance of genetic testing for patients and their families, as well as showing the genetic factors that have already been associated with increased susceptibility to the development of ASD.

Perspective

Through genetic counseling, a form of preventive medicine and improvement of health care is obtained, offering a logical explanation for the condition, especially those of genetic etiology, establishing the risks of familial recurrence, and expanding the knowledge of the disorder in order to influence its treatment and prognosis.

RESUMO

Introdução: O Transtorno do Espectro do Autismo é neurodesenvolvimental de origem multifatorial, que compromete habilidades sociais, comportamentais e de comunicação desde a infância. Com prevalência global relativamente elevada, o aconselhamento genético é essencial para compreender os fatores genéticos envolvidos e estimar o risco de novos casos na família.

Objetivo: Analisar a relevância do aconselhamento genético no contexto do autismo, focando nos fatores genéticos e no risco de recorrência do transtorno.

Método: Foi realizada revisão sistemática da literatura nas bases de dados PubMed, SciELO, BVS, Lilacs e Google Scholar, em português, inglês e espanhol. A busca incluiu os descritores "autismo", "aconselhamento genético", "genética", e "risco de recorrência", resultando na seleção de 26 artigos relevantes.

Resultado: A maioria dos casos tem origem genética, o que torna o aconselhamento genético essencial para avaliar o risco de recorrência e os fatores genéticos e ambientais envolvidos. O risco de recorrência varia de acordo com o grau de parentesco, sendo mais elevado entre parentes próximos. A hereditariedade do transtorno é mais pronunciada em gêmeos monozigóticos, e também se observa risco aumentado entre irmãos e em famílias com outros casos desse transtorno ou Transtorno de Déficit de Atenção e Hiperatividade. O Transtorno do Espectro do Autismo é mais prevalente em meninos, e o efeito materno no período pré-natal não mostra influência significativa.

Conclusão: O aconselhamento genético é crucial para esclarecer o transtorno, sendo importante tanto no setor público quanto privado, para informar as famílias sobre os riscos de recorrência e os fatores genéticos envolvidos, dado seu alto impacto na população geral

PALAVRAS-CHAVE: Transtorno do espectro autista. Aconselhamento genético. Genética.

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INTRODUCTION

Autism spectrum disorder (ASD) is a neurodevelopmental disorder that impacts social, behavioral, and communication skills, and is characterized by stereotypies, rigidity in routine, and impact on the understanding of sensory stimuli.[1] It begins in childhood and has as its etiology genetic and environmental factors that can modulate the degree of impairment of the disorder[2], which can be both idiopathic and syndromic.

These causes can be multifactorial caused by the presence of genetic variants in genes already associated with the disorder, maternal immunological and environmental factors during pregnancy, bringing autistic people impairments in social relationships, poor eye contact, lack of attention, global developmental delay and even impaired intellectual development.[3] Among the genes already associated with ASD with a high level of confidence are: ADNP, ANK2, ARID1B, ASH1L, ASXL3, CHD2, CHD8, CUL3, DSCAM, DYRK1A, GRIN2B, KATNAL2, KMT2A, KMT5B, MYT1L, NAA15, POGZ, PTEN, RELN, SCN2A, SETD5, SHANK3, SYNGAP1, TBR1, TRIP12 and UBE3A.

Palmer et al.[4] in 2017 estimated that the general population incidence of ASD is 1.25%. In Brazil, in 2023 alone, it was estimated that there are about 2 million Brazilians with autism, which represents that 1% of the population has the disorder, according to a survey carried out by the Brazilian Institute of Geography and Statistics (IBGE). In addition, it currently affects 1 in 59 children under the age of 8 according to the Developmental Disabilities Monitoring Network.[5] Based on these data, the high prevalence of the disorder is remarkable; therefore, the demand for genetic counseling services has increased, both at the request of neuropediatricians and pediatricians, and at the initiative of the family itself to obtain information and whether there is a risk of having others affected in the family nucleus.

Genetic counseling is also useful for the early diagnosis and treatment of some conditions and, as a consequence, avoid damage caused by it. Thus, this medical practice is performed by physicians with specialization in medical genetics and is applied to various diseases, and in the present study the emphasis is on genetic counseling for family members of patients with ASD.

Autism is not always easily diagnosed since it is a comorbidity of another condition. The diagnosis of idiopathic ASD is predominantly clinical; however, technological advances in genetics are providing specific genetic tests to confirm the diagnosis and, based on this, genetic counseling to understand the result is considered.

The etiology of ASD involves an important genetic factor, influenced by the segregation of variants in the family and the variation in risk, according to the degree of kinship. Family segregation is usually measured by the risk of family recurrence, which serves as an indication of a genetic factor in ASD and is variable according to the degree of kinship. Thus, it can be placed as another important factor for the realization of counseling to inform families with an autistic child about the possibility of having other children with the same disorder or another neurodivergence.[6] To provide a safe estimate of the risk of recurrence (RR) for ASD, it is necessary to perform a detailed genetic diagnosis in the child, including defining appropriate testing strategies.

A common way to estimate RRa is sibling study. The overall risk of ASD recurrence for a child with 1 or more siblings with autism has been estimated to be between 6.1% and 24.7%[4], and this prevalence is also influenced by sex. Boys are 4 times more likely to develop autism than girls. This lower incidence among girls may also be due to greater difficulty in giving the diagnosis since they better mask the symptoms of ASD.

Carter and Scherer[7] point that the RR does not influence the levels of ASD support (I, II, III); however, the sex of the autistic child has an impact on the RR, since, if the couple has an older child with autism, there is a risk of having another male child with ASD. However, if the eldest child with autism is a girl, the risk of having a second male child with autism can be as high as 21%.

Understanding the proportion in which ASD diagnoses are shared among siblings in a family contributes to the interpretation of the genetic and environmental factors that contribute to the RR of ASD, providing more effective screening and counseling and fostering closer monitoring for subsequent siblings.

In view of this, the present study aimed to show the importance of genetic counseling in autism and to analyze the risks of recurrence for family members and patients, which is a great source of concern in having more family members with the disorder, which can even manifest itself in varying degrees and associated with other neurodevelopmental comorbidities.

METHOD

The literature review was carried out with data collected from the PubMed, SciELO, VHL (Virtual Health Library), Lilacs and Google Scholar databases. The descriptors used for the research were: autism; genetic counseling; risk of recurrence, and its equivalents in English and Spanish, with AND or OR search between the years 2013 and 2024. Through this search, 5,480 articles were found, which had the title and/or abstract read, and later, 26 articles were read in full and selected for this study (Table).

DISCUSSION

ASD: Associated genes and diagnostic tests

Autism is recognized as the neurodevelopmental disorder with an inherited rate of approximately 85%.[8] In addition, approximately 10%1 of individuals with it are from families with no positive family history

TABLE — Articles selected for literature review[1-26]

Title	Authors	Year	Country	Study Type
Autism spectrum disorder in the genetics clinic: a review	Carter and Scherer[7]	2013	Canada	Literature review
Heritability of autism spectrum disorders: a meta-analysis of twin studies	Tick et al.[6]	2015	United Kingdom	Meta-analysis
Risk of psychiatric and neurodevelopmental disorders among siblings of probands with autism spectrum disorders	Jokiranta-Olkoniemi et al.[25]	2016	Finland	Cohort
association of sex with recurrence of autism spectrum disorder among siblings.	Palmer et al.[4]	2017	USA	X
Information needs in genetic testing: A needs assessment survey among Taiwanese parents of children with autism spectrum disorders	Chen et al.[18]	2018	USA	Cross-sectional study
Etiological investigation of autism spectrum disorders: State of the Art	Gonçalves, Guardino and Leão[14]	2018	Portugal	Meta-analysis
Autism. Genetic and biological aspects	Arberas and Ruggieri[3]	2019	Argentina	Meta-analysis
Recessive gene disruptions in autism spectrum disorder	Doan et al.[26]	2019	USA	X
Sibling recurrence risk and cross-aggregation of attention-deficit/hyperactivity disorder and autism spectrum disorder	Miller et al.[9]	2019	USA	X
Association of genetic and environmental factors with autism in a 5-country cohort	Bai et al.[24]	2019	Sweden	Cohort
Recurrence risk of autism in siblings and cousins: a multinational, population-based study	Hansen et al.[23]	2019	Denmark	Cohort
Genetic couseling in neurodevelopmental disorders	Blesson and Cohen[2]	2020	USA	Meta-analysis
Adaptation and validation of the Genetic Counseling Outcome Scale for autism spectrum disorders and related conditions	Yusuf et al.[17]	2020	Canada	Validation study
Factors affecting family compliance and genetic testing of children diagnosed with autism spectrum disorder	Hendel et al.[16]	2020	Israel	Cross-sectional observational study
Genetic studies and neurodevelopment of the utility of the genetic model	Calleja-Pérez et al.[10]	2020	Spain	Literature review
Genetic counseling: access for families of patients with congenital diseases	Silva[22]	2020	Brazil	Cross-sectional study
Genetic couseling as preventive intervention: toward individual specification of transgerational autism risk	Marrus et al.[8]	2021	USA	Historical cohort
Access, utilization and awareness for clinical genetic testing in autism spectrum disorder in sweden: a survey study	Hellquist and Tammimies[5]	2021	Sweden	Cross-sectional study
Parental perceptions of genetic testing for children with autism spectrum disorders	Lucas et al.[11]	2021	USA	Classical cohort
Predictors of empowerment in parents of children with autism and related neurodevelopmental disorders who are undergoing genetic testing	Peltekova et al.[15]	2021	Canada	Classical cohort
Neuroanatomy and neurophysiology in the study of autism spectrum disorders	Rodríguez et al.[19]	2021	Cuba	Meta-analysis
The pathogenesis of autism spectrum disorder	Sanabria-Barradas et al.[13]	2022	Mexico	Meta-analysis
Shared decision-making in genetic counseling: a scoping review	Chenyang et al.[20]	2022	China	Meta-analysis
Historical perspective of autism spectrum disorder	Jaramillo-Arias et al.[21]	2022	Colombia	Meta-analysis
The autism spectrum: behavioral, psychiatric and genetic associations	Genovese and Butler[1]	2023	USA	Literature review
Autism and duplication of the 17q12q21.2 region with array-CGH: a case report	Weingartner et al.[12]	2023	Brazil	Case report

for the disorder, referred to as simplex families, and are usually caused by de novo microdeletions or microduplications. On the other hand, individuals with a positive family history for the disease are called multiplex families. Research reveals that despite the significant heredity of ASD, most children with autism are children of unaffected parents.[2,8] In addition, the risk of developing ASD among children whose parents have siblings affected by the disease is 2 to 3 times higher and the occurrence among siblings is 20 times higher than in the general population, highlighting the need to recognize the genetic factors present in these families.[3]

Genetic counseling in autism

The follow-up of a geneticist for patients with autism and their families can help in several aspects, such as having an early diagnosis, assessing the risk of recurrence and understanding the genetic diagnosis of the disorder.[9] There is great concern among families about having a child with ASD when there is a family history, in which case genetic counseling is very useful to analyze this possible risk of recurrence. Thus, genetic counseling for autistic people and their families can help in understanding the diagnosis and influence the reproductive decision-making of these individuals.[9]

In autism, as in other neurodevelopmental disorders, genetic counseling can be difficult for parents and guardians of individuals with autism to accept.[10] Therefore, it is of great importance to use strategies to break stigmas that are responsible for the denial of having a child with a genetic disease, such as autism. These strategies should make them not feel guilty about the child's disorder or blame their spouses, or even be stigmatized and need help to deal with their child's condition in the best way.[11]

ASD is a condition that directly affects the social, emotional and behavioral life of the individual, compromising the construction of solid relationships and limiting the ability to interact and express themselves, with the focus often on restricted interests and repetitive acts, manifesting itself mainly in early childhood.[11,17,18] In order to minimize damage and delays in the child's integral development, an early therapeutic approach is of great importance, which should provide explanations and guidelines for an assertive multidisciplinary treatment. Most of the time, the geneticist who plays this role is the geneticist through genetic counseling.

Genetic diagnosis helps in setting realistic expectations for treatment and guiding family planning decisions, as well as allowing the person with autism to enhance their view of their own state.[11] Although, in some cases, counseling is not very enlightening for some parents or does not provide a direct benefit,[11,15] the experience of doing it still presents many gains, since it helps to empower and inform family members, who no longer acquire a passive position and start to have knowledge about the resources that enable them to gain control over the situation, collaborating in the progress of the health condition of the ASD

patient with the acquisition of quality therapies and improvement of training as caregivers.[11,15,17,18]

Risks of recurrence in ASD

The heritability of autism is described in approximately 80% of cases,[21] that is, genetics has a great influence on the development of the disorder. Jaramillo-Arias et al.[21] showed that 18% to 20% have a somatic genetic cause, that is, not hereditary, further contributing to the attention to the role of genetics in the process of understanding the disorder and its etiology.

According to a study carried out in 5 countries, in which 3 were Nordic (Denmark, Sweden and Finland) it was concluded that genetic factors explained 73.9% of the risk for developing ASD, and in 26.5% of cases they were related to gene-environment interaction.[24]

Considering that ASD is a disorder mainly of genetic cause, there are relevant data in the literature regarding the risk of recurrence of the disorder in families, with several studies that show its prevalence in siblings, twins, cousins, presenting, however, variation in family risk by the degree of kinship.[19] Thus, the determination of the empirical risk of recurrence becomes an important resource to inform families about the risks involved in presenting a relative with ASD. Therefore, estimating the risk of ASD recurrence helps for a better clinical and etiological understanding.[26]

The risk of recurrence between siblings is a possible way to assess shared genetic contributions. One way to ascertain this family risk is by analyzing siblings born after at least one child diagnosed with ASD. This type of investigation also estimates the recurrence of other neurodevelopmental diseases, such as attention deficit hyperactivity disorder (ADHD), such as if a couple has a child with ASD or ADHD and decides to have another child, they have a high chance of that second child developing any of these disorders. This estimate mentioned above reveals that the risk of ADHD is 51.92% and the risk of ASD is 48.10%, in the case of a family that already has at least one child with ASD or ADHD.[26]

Some research considers that the overall risk of ASD recurrence among siblings is between 6.1% and 24.7%.[4] However, there is an estimate regarding the risk of sex-specific recurrence. Palmer et al.[4] showed that people with autism have a RR of autism of 4.2% in female siblings and about 12.6% in males. If a woman has this disorder, the authors demonstrated that ASD was present in 7.6% of female siblings and 16.7% in males.[4]

There is a higher prevalence of ASD in males, with men being 4 times more affected than women.[11] This occurs due to the presence of a marked male bias, since in females ASD carriers have an excess of biallelic loss-of-function mutations.

Tick et al.[6] studies involving monozygotic and dizygotic twins also point to information on the heritability of ASD in these individuals; results of 74% and 93% of heritability of ASD in twins were found in the meta-analyses, and a 98% agreement in monozygotic twins against 44-60% in dizygotic twins.[3]

According to Hansen et al.[23], the concordance rate among monozygotic twins was 2 times higher than that of dizygotic twins, and the concordance rate among full siblings was up to 3 times higher than in half-siblings. The authors also point to the risk of ASD recurrence in maternal half-siblings at 5.8%, while paternal half-siblings have 3.9%, and RR in cousins at 1.9%.

The maternal effect[21] is characterized by the influence that the mother, during pregnancy, can have on the development of traits related to ASD in the child, such as characteristics or conditions that impact the child's health. This phenomenon does not have such a significant contribution to the development of the disorder, constituting a minimal association, estimated at between 0.4 and 1.6%.[21]

Regarding the influence of the shared environment, that is, particularities of the family or domestic environment that can contribute to the similarity between family members, in addition to the genetic,[21] it is not relevant to the risk of ASD.[21] However, there are several limitations that may interfere in the elaboration of this finding, given the varied genetic structure of different populations studied and analytical models sensitive to small samples.[21]

ASD, in many cases, is established simultaneously with several other disorders, such as intellectual disability, attention-deficit/hyperactivity disorder, behavioral disorders, and anxiety.[22] It is believed that the probability of acquiring psychiatric and neurodevelopmental disorders is 2 times higher[22] among siblings of autistic people, due to associated environmental or genetic factors.[22] This assumption was reinforced by a twin study, in which a dizygotic twin was diagnosed with ASD, increasing the likelihood that his brother would develop attention deficit hyperactivity disorder (ADHD), developmental coordination disorder (DCD), tics, and learning disability.[22]

Also in this study, about 36.9% of individuals with ASD had a sibling diagnosed with any psychiatric or neurodevelopment,[22] with the most prevalent risks being ASD, OCD, ADHD, learning and motor coordination deficits, as well as intellectual disability.[22] Childhood-onset disorders obtained strong associations, which were found without considering whether the child with autism had intellectual disability or not.[22] However, it is necessary to consider the divergences in studies that deal with the subject, since some of them mention a low level of cognition in the proband, others have evidenced a high level of verbal IQ, as well as there are studies reporting indifference, which reflects the wide spectrum of variation in autism.[22]

It is also assumed that childhood-onset disorders have etiological similarity and may share risk factors, with the heritability of these conditions estimated as high, in addition to environmental factors contributing to the process, as they were associated with these

disorders, in cases of prenatal infection/inflammation, obstetric or perinatal complications, and parental age.[22]

Therefore, this study highlights the importance of research regarding psychiatric and neurodevelopmental disorders among siblings of children with ASD, essential to obtain detailed and specific information about clinical, treatment, etiology, prevention and prognosis. Thus, it is of great importance that the siblings of the individual with ASD be carefully monitored by the health professional in charge. Through genetic counseling, parents should be informed about the disorder, which should have its aspects addressed in a broad way, which will contribute to the search for help, not only for the diagnosed subject, but also for their family members, who must undergo an evaluation.[4,19,22,26]

CONCLUSION

Due to the high heritability of ASD, it is coherent to use genetic counseling for this condition to bring significant information, especially in the analysis of the risks of recurrence. The overall risk of ASD recurrence among siblings is 6.1% and 24.7%, with 74% and 93% of ASD heritability in twins and 2 times higher among monozygotic twins than in dizygotic twins. In addition, there is a 4-fold higher prevalence of ASD in males and a risk of 48.10% in the case of a family that already has at least one child with ASD or ADHD. Therefore, genetic counseling to determine the empirical risk of recurrence is an important resource, if not essential, to inform families about the risks involved in presenting a family member with ASD.

Authors' contributions

João Marcos Valentim dos Santos: Preparation and initial writing
Isabela Garrett Portugal: Methodology
Marcela Sarú Campos: Methodology
Maria Müller Angulski: Study data statistics
Bruno Henrique Moraes Strapasson: Creation of models
Tailla Cristina de Oliveira: Critical review
Liya Regina Mikami: Critical review and final writing

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