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Follow-up of patients with Autistic Spectrum Disorder and Down Syndrome in a medical genetics outpatient clinic: experience report

Acompanhamento de pacientes com Transtorno do Espectro Autista e Síndrome de Down em ambulatório de genética médica: relato de experiência

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Abstract

Early diagnosis of Autistic Spectrum Disorder (ASD) and/or Down Syndrome (DS) in children and their outpatient follow-up are essential for their development and maintenance of quality of life. The objective of this study is to report the experience of students of the Academic League of Medical Genetics in the outpatient follow-up of these patients in the city of Olinda, Pernambuco, between March and November 2022. The students were able to improve their care, especially in Medical Genetics, making it more humanitarian and comprehensive, due to the growing demand of these patients who, due to their comorbidities, need to be referred to other specialties, in order to provide regular and specialized follow-up.

Keywords: Autism Spectrum Disorder, Trisomy 21, Heredity.

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Resumo

O diagnóstico precoce do Transtorno do Espectro Autista (TEA) e/ou Síndrome de Down (SD) em crianças e o seu acompanhamento ambulatorial são essenciais para o seu desenvolvimento e manutenção da qualidade de vida. O objetivo deste trabalho é relatar a experiência dos discentes da Liga Acadêmica de Genética Médica no acompanhamento ambulatorial desses pacientes no Município de Olinda, Pernambuco, no período entre março e novembro de 2022. Os ligantes puderam aperfeiçoar o seu atendimento, especialmente na Genética Médica, tornando-o mais humanitário e integral, devido à crescente demanda desses pacientes que necessitam, pelas suas comorbidades, do encaminhamento para outras especialidades, de forma a proporcionar um acompanhamento regular e especializado.

Palavras-chave: Transtorno do Espectro Autista, Trissomia do 21, Hereditariedade.

INTRODUCTION

Autism spectrum disorder (ASD) is a multifactorial condition related to genetic, environmental, immunological, and neurological factors, but with no pathognomonic sign, hindering diagnosis.[1] In this sense, ASD is related to the behavior and the difficulties in normal social interaction, intensification of standardized repetition, and sometimes aggressive attitudes.[2]

Thus, it is fundamental to dedicate attention to the service of children with ASD because the number of diagnosed cases increased. For this reason, the expansion of follow-up outpatient and their criteria for a safe diagnosis, which is based on clinical aspects legitimized by the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV).[1]

Down Syndrome (DS) is a more common chromosomal disorder related to intellectual deficits in the neurodevelopment of the child, such as difficulties in learning and communicating, and long-term complications (e.g., heart disease, visual problems, and endocrinopathies). The early treatment at the follow-up of these children often includes therapies, such as physical therapy and speech therapy, which are essential for adequate growth and promoting the maintenance of their quality of life.[3] Therefore, children with DS live with several barriers to specialized care.[4]

Thus, it is necessary to have adequate follow-up since these populations require targeted attention, mainly due to the demands caused by comorbidities, which impair communication and neurodevelopment in the long term. Therefore, medical genetics outpatient is fundamental for mitigating the harmful effects of these conditions.[5]

METHODS

This experience report described the follow-up of patients with ASD and DS performed by students from the Medical Genetics League, under the supervision of the geneticist, during their outpatient services from March 2022 to November 2022.

The data collection was based on discussions with the genetics preceptor doctor from the clinic and the biweekly meetings of the Medical Genetics League aimed at ASD and DS.

RESULTS

The follow-up began with the emergence of genetics outpatient focused on children and adolescents with ASD and DS aged over 16 years. To better organize this activity during all follow--up periods, the students were divided to ensure the presence of at least one of them at each turn to achieve the greatest number of medical appointments. Meetings were directed at students, focusing on cases, seminars, and the genetic mechanisms related to these conditions.

Besides the knowledge obtained in the meetings, the responsible geneticist explained these topics, emphasizing the medical genetics outpatient clinic of this specialty. Questions related to the anamnesis were also addressed to help confirm the diagnosis, such as food aspects, triggers factors of irritability and aggressiveness, practice in physical activity, and family relationships. Furthermore, the main prescribed medications were exposed, aiming to treat certain signs and symptoms, such as difficulty concentrating and learning, and aggressive behavior; the latter is most common in patients with ASD. The most used medication for aggressive behavior is risperidone, an antipsychotic that blocks serotonin and may cause adverse effects in the long term, such as antipsychotic-induced weight gain, metabolic syndrome, and hyperprolactinemia.[6]

During the appointments, the students had the opportunity to perform the physical examination and distract the children with the toys of the clinic, while their parents were in the medical appointment. Despite their simplicity, these actions made it possible to improve the medical care and perspective of students, mainly in the completeness and equity of the health service of patients with ASD and DS, respecting their particularities.

CONCLUSION

The genetics outpatient clinic may be beneficial for students, especially at the follow-up of patients with ASD and DS due to their growing demand. Moreover, these patients present comorbidities that requires genetics support, other specialties.

Although it is not a compulsory internship, the genetics outpatient clinic becomes an important complementary teaching tool for students. The experience improves their perception of integral care for patients with ASD and DS, making them future professionals more humanitarian.

CONFLICT IN INTERESTS

None.

CONTRIBUTIONS OF THE AUTHORS

TJMBS: preparation of the abstract, introduction, experience report, and final considerations; **HEEV** and **ANF**: preparation of the abstract, introduction, experience report, and final considerations; **HCPO** and **LLRS**: preparation of the introduction, experience report, final considerations, and references formatting; **AESM** and **PSCA**: guidance, correction of the writing and approval of the final version. All the authors approved the final version of the report.

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