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SEVERE COMBINED IMMUNODEFICIENCY: THE IMPORTANCE OF EARLY DIAGNOSIS IN THE PEDIATRIC POPULATION

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Key words: "Early Diagnosis", "Severe Combined Immunodeficiency" and "Newborn Screening".

INTRODUCTION

Severe combined immunodeficiencies (SCID) are hereditary diseases characterized by a deficient immune response. Furthermore, they are responsible for high morbidity and mortality in pediatric patients and, therefore, it is essential to early evaluate newborns through preventive actions, such as neonatal screening, in order to obtain a diagnosis as quickly as possible, aiming for better results. long-term results and, consequently, a good prognosis after treatment.

GOALS

To understand the need for early diagnosis of severe combined immunodeficiency in the pediatric population to obtain adequate treatment and prognosis.

METHODOLOGY

Literature review, with a qualitative approach, based on the databases Scielo, Revista Brazilian Journals, Revista Brasileira de Alergia e Immunologia. Descriptors such as "Severe Combined Immunodeficiency", "Early Diagnosis" and "Newborn Screening" were used. For the inclusion criteria, articles published in the last 10 years, in Portuguese, were selected. Exclusion criteria were based on the timeliness and level of evidence of the studies. In the end, it was restricted to 4 articles.

RESULTS AND DISCUSSION

SCID is characterized by an alteration in the immunological response composed of T lymphocytes and/or B lymphocytes and/or NK cells, generating an increased vulnerability to infections. The diagnosis is based on a complete and detailed history, physical examination and laboratory tests, and treatment is preferably carried out by bone marrow transplantation. Furthermore, this pathology is considered a pediatric emergency due to the high rate of morbidity and mortality in the first year of children's lives and its late diagnosis corroborates the high death rates. Furthermore, the treatment, when applied before four years of age, has a 95% survival rate, while if carried out after this period, the rate drops to 60% and the incidence of complications and sequelae increases significantly. The best tool to increase the incidence of early diagnosis and, therefore, obtain more efficient treatment is neonatal screening. SCID screening is done by quantifying markers of the normal development of T lymphocytes, called T cell receptor incision circles (TRECs), which are shown to be in low numbers or identifiable quantities. For better diagnostic accuracy, IL-7 is measured simultaneously, as this is related to the adequate development of T cells. The combination of these two tests allows a diagnostic sensitivity of approximately 100%. Studies on the efficiency of neonatal screening showed an increase in survival, showing that around 85% of those tested at birth survived, while only around 58% survived when not tested. In Brazil, there are neonatal screening tests for more than 38 diseases, but only six are included in the free test offered by the SUS, popularly known as the "Feet test". Primary immunodeficiencies, such as SCID, are in the group of screened diseases that are not offered free of charge.

CONCLUSION

Therefore, it is important to understand the need for an improvement in neonatal screening, as its efficiency, combined with a detailed anamnesis and complete exams, are decisive in the early diagnosis of Severe Combined Immunodeficiencies, influencing the prognosis.

REFERENCES

TROMBINI, Ana Beatriz Dalla Déa; GUIRALDELI, Anna Julia; RIBEIRO, Lucas Gaspar. A importância da inclusão da imunodeficiência combinada severa na triagem neonatal pública do Brasil. **Brazilian Journal of Development**, v. 6, n. 10, p. 79906-79909, 2020.

PFISTERER, Juliana Cantagalli et al. Imunodeficiência combinada grave: uma revisão da literatura. **Arquivos de Asma, Alergia e Imunologia**, v. 2, n. 2, p. 56-65, 2014.

KANEGAE, Marília PP et al. Triagem neonatal para imunodeficiência combinada grave. **Rev bras alerg imunopatol**, v. 34, p. 7-11, 2011.

RIZZO, Luiz Vicente. DIAGNÓSTICO PRECOCE DAS IMUNODEFICIÊNCIAS COMBINADAS GRAVES. **Revista Paulista de Pediatria**, v. 35, n. 1, p. 2-2, 2017.