

Natureza do trabalho: Artigo de Revisão

TÍTULO

RETT SYNDROME, THE ROLE OF MECP2 IN AUTISM — AN OVERALL REVIEW

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RESUMO

Introduction: The Rett Syndrome (RTT) was first described by Dr. Andreas Rett in 1966 in German (10). It is a progressive neurodegenerative disorder, which causes mental retardation, seizures and the carrier also present autistic behavior. It's a genetic disorder since its inheritance pattern is due to the X chromosome, technically named X-linked dominant. Consequently, the mostly affected individuals are females — the statistics shows that 1 in 10.000 - 15.000 is affected by this syndrome, and that more than 95% are de novo mutations. **Objectives:** To present an overall view about Rett Syndrome, the responsible gene and its mechanisms, the diagnosis and prognosis, phenotype and symptoms, inheritance patterns and possible treatments and therapies. **Material and Methods:** The consulted papers were obtained at PubMed Central® and Web of Sciece. Search key terms included RTT, MECP2, autism, Rett Syndrome, de novo mutations. **Discussion and Conclusion:** RTT is a neurodegenerative disorder responsible for an autism spectrum and its development is separated in 4 stages: The first stage (stage 1) is named Stagnation Stage and it occurs between 6 and 18 months, and it's noticed a developmental delay in relation to healthy kids. The second stage (stage 2) is called Regression Stage and occurs from 1 to 4 years old, it is when the autistic behavior becomes perceivable and when happens a regression of acquired skills. The third stage (stage 3) is the pseudo-stationary phase when the loss of skills stops and hand features starts to manifest (gait dyspraxia and ataxia). The fourth and last stage (stage 4) is the late motor decline phase, it is inadequately defined as complete loss of the ability to walk and it happens around the twenties, it may also cause Parkinson's disease similar symptoms. It is considered inadequately because some of RTT patients might have never learnt how to walk (during stages between 2 to 3). **Discussion and Conclusion:** RTT is not yet curable but currently researches are leading to a greater view about this syndrome. Based on that we can be optimistic about a future therapy that will be able to revert symptoms if early diagnosed. Thus, we have to know the syndrome characteristics, the origin, the mutations spectrum, the MECP2 function and location and how RTT patients' cells react to experimental processes. Scientists have yet to investigate better approaches based on similar inheritance patterns of different.

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