中南大学学报(医学版) 2011, 36(8) 703-711 DOI: 10.3969/j.issn.1672-7347.2011.08.001 ISSN: 1672-7347 CN: 43-1427/R

精神病学与精神卫生专栏

孤独症遗传学

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摘要：

孤独症是一组具有病因和临床异质性的神经发育性疾病,通常发病于3岁以前。孤独症具有3个典型的核心症状:语言交流缺陷,社交障碍以及狭隘兴趣和重复的行为。孤独症发病率在全球范围内呈增长趋势。双生子和家族聚集性研究发现遗传因素在孤独症的发病机制中起重要作用(遗传度>90%)。遗传学研究发现了孤独症的一些易感基因和位点,但仍然有70%~80%的孤独症患者遗传病因不明。

关键词： 孤独症 异质性 表观遗传学 拷贝数变异

Genetics of autism spectrum disorders

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Abstract:

Autism is a group of etiology and clinical heterogeneous neurodevelopmental disorders with an onset before 3 years old. It has 3 core characteristics: deficits in verbal communication; impairment of social interaction; restricted interests and repetitive behaviors. The incidence is increasing over time worldwide. Twin and family studies have demonstrated that autism has a high heritability (>90%). Although certain progress of autism genetic study has been made in the last de-cades and several autism susceptibility genes and loci have been identified, there are still about 70 %- 80% of patients for whom an autism-related genetic change cannot be identified.

Keywords: autism heterogenicity epigenetics copy number variation

收稿日期 2011-06-20 修回日期 网络版发布日期

DOI: 10.3969/j.issn.1672-7347.2011.08.001

基金项目:

This work was supported by Scholarship Award for Excellent Doctoral Student granted by Ministry of Education of China and National Basic Research Program of China (2010CB529601 & 2012CB517902).

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