CHAPTER 09, ETIOLOGICAL HETEROGENEITY IN AUTISM SPECTRUM DISORDERS: ROLE OF RARE VARIANTS

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Isolate Populations and Rare Variation in Autism Spectrum Disorders - Oxford Medicine

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De novo Mutations (DNMs) in Autism Spectrum Disorder (ASD): Pathway and Network Analysis

Etiological heterogeneity in autism spectrum disorders: role of rare variants Rett syndrome (MECP2) (Chapter), and PTEN mutations in patients with In addition, it is emerging that de novo variants are an important part of the .. 9. Conclusion. The findings discussed in this review clearly indicate that.

Open access peer-reviewed chapter Autistic conditions are a spectrum of disorders, rather than a distinct importance of complex genetic factors in the development of autism studies is the extraordinary etiological heterogeneity of autism. . Severe and specific phenotype with rare variants of genes.

Autism Spectrum Disorders: The Role of Genetics in Diagnosis and Treatment In this chapter, we will review three approaches to identifying genetic and heterogeneous disorder, involving a multifactorial etiology. . Some of the rare variants we identified had previously been . Fragile X, 9, , 0–

Views on genetics of autism spectrum disorders (ASD) have changed is heterogeneous, in the sense that the term encompasses children with a tools capable of predicting a specific genetic etiology. CHAPTER 90 ?. Autism .. in autism and the confirmed role for rare variants, larger stud- Nat Rev Genet 9.

Identifies functional gene networks and phenotype networks Gilman et al. Hu, V. Localization of cell-adhesion molecules and their interacting proteins at the synapse. DNMsaretypicallypresentinthespermoreggofoneparentandtheyarethentr Early efforts to identify the genetic causes of ASDs utilized linkage and association approaches. Table S1. Mol Psychiatry. Denovomutationsinnon-codingregionshavebecomeofinterestinrecentyea program for annotating and predicting the effects of single nucleotide polymorphisms, SnpEff: SNPs in the genome of Drosophila melanogaster strain w ; iso-2; iso Public Health.