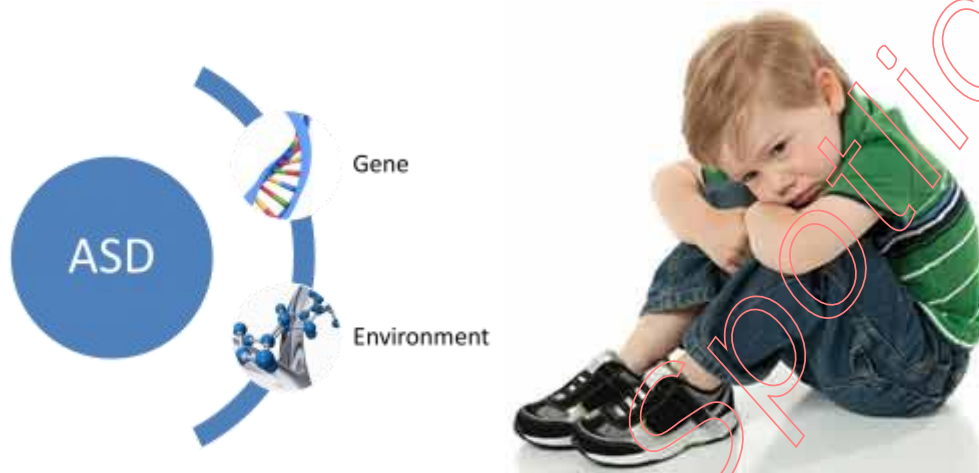


Genetic Science Spotlight

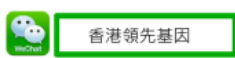
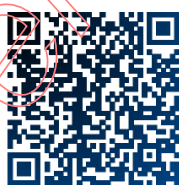
Genetic Counseling as a Key Elements of Comprehensive Gene-Environment Understanding for Individual ASD Families



Autism Spectrum Disorder (ASD) is a multifactorial disease, influenced by genetics, environment before and after birth, as well as gene-environmental interactions. Despite merely 10% of cases can be identified with a pathogenic gene mutation, public health statistics suggests that influence of inheritance in ASD is indisputable. When all testing in the proband is negative or inconclusive, the recurrence rate (RR) is extrapolated from empirical studies. Retrospective family studies published prior to the late 1990s estimated the RR for a couple with one autistic child to be 3–10%. A recent large (n = 664), longitudinal, prospective study of high-risk infants (younger siblings of proband with ASD) has shown that the recurrence rate is higher than previously estimated, as 18.7% of ‘baby sibs’ were ultimately diagnosed with an ASD. The RR did not change with the severity of ASD symptoms or with the sex of the proband. The sex of the next child, however, does impact on RR, as males are consistently at higher risk for ASDs. When two siblings in one family are affected with ASD, the RR for younger sibs increased to 32.2%, which is consistent with older estimates of 25–35. Provided with genetic counseling, ASD families can hence better understand their next steps and/or obtain referral to get help from the right specialists.

Carter, M. T., Scherer, S. W. (2013). Autism spectrum disorder in the genetics clinic: a review. Clin Genet 2013. doi: 10.1111/cge.12101

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