

Genetic Science Spotlight

Association of mutation in non-coding region related to autism in the recent biggest study to-date by MSSNG incites a need for whole genome sequencing



The study has explored in an unprecedented manner in technological methods and numbers of genome sequenced to further deepen the understanding of genetics in autism. The data generated by MSSNG, a collaboration between Autism Speaks and Sick-kids are solely from de novo cases with unaffected parents, or known as simplex cases. Sequencing of 600 genomes with phenotypes systematically filed represent the largest database to date.

Discovery of differences of mutation inherited from mother and father was observed. Increasing age of fathers are more likely to cause autism-related mutation in offspring whereas mothers are more likely to cause clustered/concentrated stretches of mutation usually near copy number variant site. This spurred researchers to believe there might be differences in DNA repairing system in men and women.

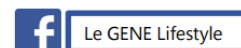
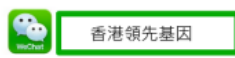
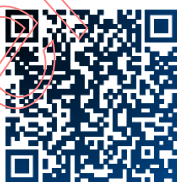
Those mentioned so far are germline mutation, however, it has also been found from this study that mutation that occur immediately after fertilization take up 1% of the 600 genome studied. This early embryonic mutation are caused by environmental insults that disrupt the normal epigenetics. Environmental insults could be from pollutants, nutritional deficiencies and inflammation.

Furthermore, there are mutation in genes that cause cascading effect in other genes that is responsible of brain development. This cascading effect explained the repeating series of genes mutation that are widely observed.

This landmark study is geared to improve diagnoses and precision treatment. Given the more in depth understanding in ASD genetics, patients and their doctors can benefit from the insights that genetic counselors have to offer.

<http://www.nature.com/articles/npjgenmed201627>

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