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Segmental duplications in neurodevelopmental, neurological and behavioral disorders

Status	Past
Competition	Genoma España/Genome Canada
Sector	Health
Genome Centre	Ontario Genomics Institute
Project Leaders	Stephen Scherer (Canada) & Xavier Estivill (Spain)

Project Description

Genetics is known to contribute significantly to many of the neurodevelopmental and behavioral diseases but in most cases, the causative molecular defect has not yet been determined. Therefore, there are no tests for early detection and diagnosis of these disorders and there is little information pertaining to their biological basis. An increasing number of neurological and behavioral disorders, however, are due to changes in the architecture of specific sites along the DNA of chromosomes in the human genome.

The importance of such chromosome alterations has further been stressed with the information obtained with the human genome sequence, which has now been shown to harbor "molecular signatures" which facilitate rearrangements of genomic material. Furthermore, it has been shown that there is variability among individuals and species regarding the genomic structure and the copy number of genes located at specific regions of the genome, which, in turn, seem to play an important role in evolution.

The regions, that comprise an amazing 5% (150 million chemical bases) of the content of the human genome and contain these molecular signatures, known as segmental duplications or duplicons, have not yet been accurately or completely characterized by the large-scale DNA sequencing projects.

Other principal investigators include scientific research groups from the University of Toronto, the University of British Columbia, SeeDNA Biotech, Fundacio Parc de Recerca Ciomedica de Barcelona, Universidad Pompeu Fabra, MedPlant Genetics (Spain), and CAGT-Citogen (Spain).

A SUMMARY OF THE OUTCOMES FROM THIS COMPLETED PROJECT IS CURRENTLY BEING CONSTRUCTED. PLEASE CHECK THIS SITE AGAIN FOR AN UPDATE ON PROJECT RESULTS.