Identification of the molecular mechanisms in Rett syndrome and related disorders

Acronym: RTT-GENET
Organisation: Bilkent University

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Applicable Instrument: Integrated Project

Sub-Thematic Priority most relevant to your topic:
1.1.1.ii Combating major diseases

Other relevant Sub-Thematic Priorities:
1.1.1.i Advanced genomics and its applications for health

Abstract:
Rett syndrome is a severe neurodevelopmental disorder that arises from de novo mutations in the X-linked MECP2 gene. This project is devoted to clarify the molecular mechanisms associated with Rett syndrome and related phenotypes. Specific aims are:
2. Identification of MECP2 target genes; assessment of their role in neurological disorders.
3. Identification of modifier genes in MECP2-mutated patients.
5. Identification of molecular mechanisms and genes that are associated with CpG dinucleotide hypermutability.
6. Collaboration with foundations, associations to improve management of patients.

Additional document: EOI_RTT-GENET_V1.doc

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