



Identifying genes involved with aneuploidy syndromes has proven difficult. Possible reasons are allelic variability, and genetic interactions between these variable alleles and the genetic defects that are the underlying cause of the disease. To unravel the relationship between genotype and phenotype, large patient cohorts are needed.

The AnEUploidy Project aims to collect a large number of patients with rare chromosome 21 anomalies. These patients will be phenotyped according to a standardized phenotypic list and genotyped with array analysis (targeted chromosome 21 array of Nimblegen®) (no costs) . The cytogenetic and clinical data will be stored in two existing databases, ECARUCA and ANEU21.

If you have diagnosed a patient with partial trisomy 21 or monosomy 21 and would appreciate further study with array analysis the procedure is the following:

1. After informing us about the patient we will send you a phenotypic list to collect the clinical details and a consent form with an information letter for the parents.
2. 10 cc Heparin blood (for cell line) and/or 10 µg of DNA will be needed to perform array analysis.
3. We will correspond the outcome of the array analysis with you.

This study will be performed in collaboration with different European centres.

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