



European 6th framework project ANEUPLOIDY

Aneuploidy is related to gene expression perturbation and abnormalities, but the molecular pathogenesis of the numerous aneuploid disorders is largely unknown.

One of the aims of the AnEUploidy project is to collect a large number of patients with clinically defined syndromes. These patients will be phenotyped according to a standardized phenotypic list and genotyped with the Affymetrix® Genome-Wide Human 6.0 SNP Array featuring 1.8 million markers to see whether a underlying microdeletion or duplication is present.

The study will, initially, focus on the following syndromes:

1. Ohdo
2. Baraitser-Winter
3. Coffin Siris
4. Hallermann-Streiff
5. Malpuech
6. Schinzel Giedion
7. Floating Harbor
8. IFAP
9. Aase-Smith
10. Bohring

If you have patients who might have one of these syndromes and you are not sure about the diagnosis you may email as well for a clinical opinion by one of our experts.

If you have patients with one of the above named syndromes and would appreciate further study with SNP array (no costs) the procedure is as follows:

- After you inform 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.
- 10 µg of DNA will be needed to perform the array analysis.
- We will correspond the outcome of the SNP array analysis with you.

For further details and/or questions please contact:

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