



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 to see whether a underlying microdeletion or duplication is present. Besides we provide whole exome sequencing (SOLiD, life technologies).

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 (solved¹: clinical follow-up study)
7. Floating Harbor
8. Cantu syndrome
9. Aase-Smith
10. Bohring

If you have patients with one of the above named syndromes and would appreciate further study (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.
- 30 µg of patient DNA and parental DNA will be needed to perform the array analysis and whole exome sequencing. Preferably also include a cell line.
- We will correspond the outcome of the analyses with you.

For further details and/or questions please contact:

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¹ Hoischen A., van Bon B.W., Gilissen C., et al. De novo mutations of SETBP1 cause Schinzel-Giedion syndrome. Nat Genet 2010;42(6):483-5