

## Research Page for chromosome 18q deletions



We propose to do a detailed genotype-phenotype analysis within the ECARUCA framework on patients with 18q deletions.

Most of these patients carry a terminal 18q deletion, also named de Grouchy syndrome, which is characterized by short stature, congenital aural atresia, foot deformities, white matter abnormalities on brain MRI and mild to moderate mental retardation.

Another, less frequently encountered form of 18q deletions is a proximal interstitial deletion. Proximally located interstitial deletions mainly lead to microcephaly, short stature, cleft lip/palate and severe mental retardation. The existence or absence of clinical features depends on the size and position of the deleted region, although patients in whom the breakpoint is localized within the same chromosome band can show many differences in phenotype.

Within our centre in Nijmegen, we have collected the clinical data and DNA samples of a cohort of 45 patients with 18q deletions. All deletions are determined by routine karyotyping and more than half of these are fine mapped by a chromosome 18 specific array CGH. This enabled us to produce an updated genotype-phenotype map<sup>1</sup>.

Continuation of investigation of patients with (sub) microscopic deletions of 18q could lead to a further delineation of critical regions and the elucidation of the molecular background of the syndrome.

We would like to propose a **collaborative study**, in which all ECARUCA account holders can enter the following patients:

- Patients with an 18q deletion known in your centre.
- Patients with an 18q deletion phenotype<sup>1</sup> in whom no deletion has been detected yet.

### What you need to do:

- Collect clinical details for all patients by use of a standard questionnaire.
- Obtain informed consent from the patient for participation in the study.

### What is done in the Nijmegen centre:

- We offer DNA typing to establish the exact size of the deletion. This work will be done using a tiling path array CGH or DNA analysis.

### Publication/dissemination of results:

- In case a scientific article is established, co-authorship is offered.

1. Feenstra I, Vissers LE, Orsel M, van Kessel AG, Brunner HG, Veltman JA, van Ravenswaaij-Arts CM. Genotype-phenotype mapping of chromosome 18q deletions by high-resolution array CGH: an update of the phenotypic map. *Am J Med Genet A*. 2007;143(16):1858-67.

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