Subtelomeric 1q Deletion Research Page

Submicroscopic 1qter deletions, including distal deletions of 1q43 or q44 → qter, lead to a recognizable phenotype, which includes mental retardation, distinct facial features, growth retardation (prenatal onset), severe progressive microcephaly, seizures and various midline related defects such as agenesis/hypoplasia of the corpus callosum, cardiac anomalies, and genital and gastro oesophageal abnormalities.

Within our group, we have collected the clinical data and DNA samples of a cohort of 13 patients with a submicroscopic 1qter deletion. The clinical presentation of these patients has clear similarities with previously reported cases with a terminal 1q deletion. However, the number of patients with a submicroscopic 1q44 deletion is relatively small, which hampers the delineation of the clinical phenotype and the elucidation of the molecular background of the syndrome.

In order to do a thorough clinical analysis of patients and to identify the gene(s) responsible for the phenotype we would like to propose the following collaborative study:

1. We would like to be informed on the numbers of subtelomeric 1q deletions (and duplications!) that you have diagnosed so far, and whether these have been published.
2. Clinical details, DNA and a cell line of these specific cases will be collected.
3. We offer DNA typing to establish the exact size of the deletion or duplication and future expression studies of interesting candidate genes.

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Reference:

Van Bon et al. Clinical and Molecular Characteristics of 1qter Syndrome: Delineating a Critical Region for corpus callosum agenesis/hypogenesis. J. Med. Genet. 4 jan 2008. [Epub ahead of print]

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