Subtelomeric 9q Deletion Research Page

The clinically recognizable 9q subtelomeric deletion (9q-) syndrome comprises severe mental retardation, hypotonia, brachycephaly, flat face with hypertelorism, synophrys, anteverted nares, cupid bow or tented upper lip, everted lower lip, prognathism, macroglossia, conotruncal heart defects and behavioural problems. In a recent study, we have shown that haploinsufficiency of the *EHMT1* gene is responsible for the main features of this 9q- syndrome[1].

In order to further study the 9q telomeric region, we are interested in patients with (interstitial) telomeric 9q deletions or duplications, whether are not encompassing the *EHMT1* gene. We will investigate if and to which extend, other genes in this region contribute to the 9q- phenotype.

We suggest the following **collaborative study**:

1. We would like to be informed on the numbers of subtelomeric 9q deletions (and duplications!) that you have diagnosed.
2. We would like to be informed about the clinical details for these specific cases
3. We offer DNA typing to establish the exact size of the deletion or duplication.

Contact person

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1. Kleefstra et al. Loss-of-Function Mutations in *Euchromatin Histone Methyl Transferase 1* (*EHMT1*) Cause the 9q34 Subtelomeric Deletion Syndrome. 2006; 79, pages 370–377

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