Subclinical features of cleft lip and palate in non-affected parents of individuals with cleft lip and palate in relation to their genetic background

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Background
Clefts of the lip (fig.1) and / or palate (fig.2) are birth defects that are caused by an abnormality in the early development of the face and occur when separate areas of the upper lip and roof of the mouth do not join together properly during development. The genetic basis of cleft lip and / or palate however remains obscure.

Methodology
This will be a population based case - control study (40 cases and 40 controls) conducted over 9-12 months. We will compare the following in parents with children with cleft lip and / or palate, with a group of volunteers who do not have any history of cleft lip and / or palate in their families:
1. Features of the lips (such as lip whorl pattern) obtained from lip prints (fig.3), and features in the superior Orbicularis Oris muscle (fig.4) obtained using ultrasonography.
2. Teeth and arch dimensions.
3. Facial characteristics using three dimensional (3D) photography (fig.5) for the face.
4. Genetic material will be obtained. This will enable a comparison of the phenotypic features of parents versus controls, and simultaneous DNA analysis of a range of candidate genes will be conducted (fig.6).

The participants will be of Celtic background with an absence of microform / overt features of a cleft. The controls will have no history of a cleft in three generations and will be older than 19 years of age.

Aims
To identify the association between the features of the lips, teeth and facial shape and specific genes in the parents of children with clefts in comparison to a sample of control volunteers, and to improve clinician ability to estimate risk of a further child having a cleft of the lip with or without cleft palate.

Analysis of Data
- Candidate genes analysis in cases and controls.
- Genotype / Phenotype correlation in cleft cases.
- Phenotype in relation to cleft type.

References
1- MARAZITA, M. L. 2007. Subclinical features in non-syndromic cleft lip with or without cleft palate (CL/P); review of the evidence that subepithelial orbicularis oris muscle defects are part of an expanded phenotype for CL/P. Orthod Craniofac Res, 10, 85-7.