More than 125 FOXL2 mutations and variants in BPES and POF patients in the Human FOXL2 Allelic Variant Database. (meeting abstract)

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Blepharophimosis-ptosis-epicanthus inversus syndrome (BPES, MIM 110100) is an autosomal dominant genetic condition in which an eyelid malformation is associated (type I) or not (type II) with premature ovarian failure (POF). In 2001, mutations in the FOXL2 gene, encoding a forkhead transcription factor, have been shown to cause BPES type I and II. Since then a number of publications appeared describing FOXL2 mutations in BPES patients. In addition, there have been reported a few FOXL2 variants in POF patients and XX males. Previously, our group has reported the existence of two mutational hotspots in FOXL2 and of intra- and interfamilial phenotypic variability in BPES families. Moreover, we have demonstrated genotype-phenotype correlations for a number of mutations in BPES patients. Here we describe a new locus-specific Human FOXL2 Allelic Variant Database (http://allserv.ugent.be/~jvdesomp/foxl2/), created using the MuStaR software (on which the PAX6, PAX2, SHOX and MLYCD Allelic Variant Databases have been based similarly). Our database contains general information about the FOXL2 gene, as well as details about more than 125 intragenic mutations and variants of FOXL2, obtained from published papers and abstracts of meetings, and also from unpublished data of our group. Not included in the current version of the database are complete FOXL2 deletions, microdeletions and cytogenetic rearrangements of the FOXL2 region on 3q23.
The aim of this database is to provide an online resource, allowing remote users to do queries by selecting options on a web form and to submit new mutations to the database by means of a submission form. We believe this database will be very useful as it contains prevalence data about disease-causing mutations, a catalogue of polymorphisms and as it will facilitate more accurate genotype-phenotype correlations to be made.