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intragenic mutations and variants of *FOXL2*, obtained from published papers and abstracts of meetings, and also from unpublished data of our group.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.