

Study of Genetics of Human Disorders of Sexual Development. SCOPEs 2013-2016: Joint Research Projects

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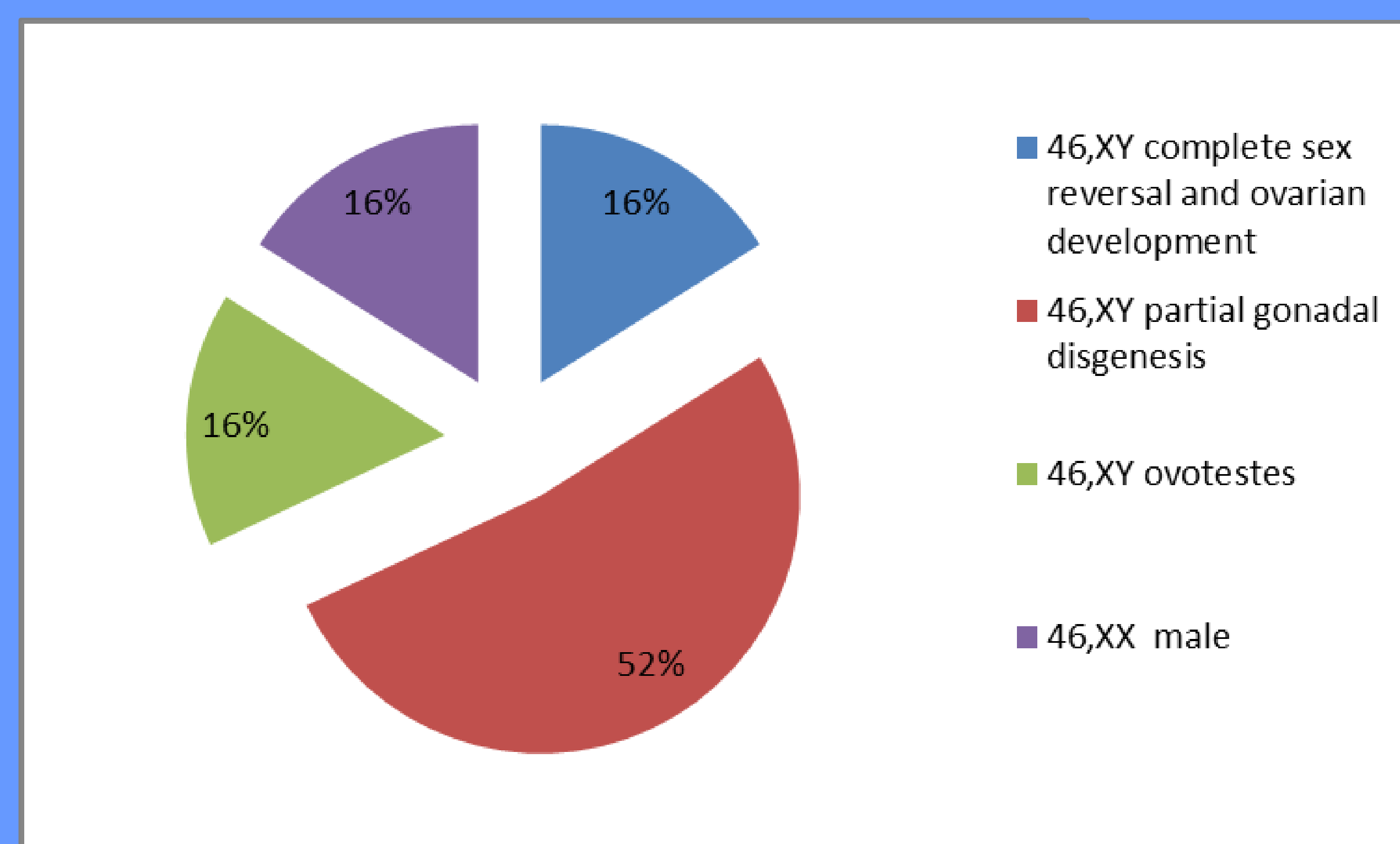
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Objectives:

Disorders of sex development (DSD) are a group of congenital developmental disorders in which the chromosomal, gonadal, or anatomical sex is atypical. The clinical diagnosis and management of DSD are difficult and complex because of the various aetiology and diverse manifestation. The project “Genetics of Human Disorders of Sexual Development” is funded by Swiss National Science Foundation and fulfilled by the University of Geneva Medical School (Switzerland), the Medical Centers from Armenia, Poland and Ukraine. The goal is to identify mutations underlying unresolved DSD phenotypes – in novel DSD genes, or regulatory regions that lead to atypical gene expression.

Methods:

Identification of new genes involved in human sex determination and differentiation is carried out through exome sequencing and CGH microarray in parallel. Armenian partner is participating in all stages preceding the exome sequencing: clinical data collecting (caryotype, family history, physical examination, ultrasound, hormonal status, surgery, histology), caryotyping, DNA samplings (proband, parents, siblings), SRY gene deletion detecting and SRY, SOX9, WT1, SF1, LHX9, RSPO1, FOXL2, WNT4, DMRT1, DMRT2 genes Sanger sequencing.



Results:

During 2 years Armenian part collected 25 DNA samples from 25 DSD cases – 4 patients with 46,XY complete sex reversal and ovarian development, 13 patients with 46XY partial gonadal dysgenesis, 4 patients with ovotestes and 4 of 46,XX DSD male. We also collected DNA samples from parents.

Conclusions:

We expect the research will provide the opportunity to develop new genetic tests for DSD diagnosis and to improve understanding of the molecular mechanisms of ovarian and testicular differentiation. The results on the cohort will be discussed in details.

References:

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