



## Details of Study ID 202305\_SM

<b>Name of the Principal Investigator:</b> Margherita Sepich
<b>Contact details including institution:</b> Paediatrics Unit - University of Pisa, Pisa, Italy m.sepich@studenti.unipi.it
<b>Coinvestigators:</b> Faisal Ahmed, Glasgow, Silvano Bertelloni, Pisa, Nina Tyutyusheva, Pisa, Angela Lucas-Herald, Glasgow, Inas Mazen, Cairo, Martine Cools, Ghent, Junfen Fu, Zhejiang, Şükran Poyrazoğlu, Istanbul, Olaf Hiort, Luebeck.
<b>Date of Approval:</b> May 2023
<b>Name of study:</b> Gonadal function and outcome in 46, XX testicular/ovotesticular DSD
<b>Summary of the proposed work to be performed with the registry data:</b> 46, XX testicular and 46, XX ovotesticular disorders of sex development are relatively rare. Presently no description of the clinical course and gonadal function has been performed in people with these conditions. This study will analyse specific clinical, biochemical, radiological and genetic features at birth, through childhood and after puberty to consider the outcome of people with these conditions. Information will also be gathered to determine whether there are any differences which may help in the differential diagnosis of these disorders, particular as they may be part of a continuous spectrum of a single genetic entity.
<b>Lay summary for the public (for the I-DSD/CAH websites and other publicity materials) (maximum 50 words)</b> This study will seek to outline the clinical course and outcome of people with 46, XX testicular and 46, XX ovotesticular DSD a group of very rare conditions. The study will allow us to understand the natural history of this group of disorders and we expect that the results will lead to improved care of people with these conditions.
<b>Inclusion criteria</b> Specific diagnosis of 46, XX testicular and 46, XX ovotesticular DSD
<b>Exclusion criteria</b> All other forms of DSD.



**Data to be collected for all participating cases:**

*Core:*

Consent to share data (Y); Centre; Country; Date of Birth; Sex at birth; Country at birth; Birth weight; Birth length; Birth head circumference; Gestational age; Current gender; Karyotype (46,XX); Disorder type (Disorder of gonadal development); Actual diagnosis (Testicular DSD/OT DSD); Associated conditions; Date of condition onset; Date of diagnosis; Associated conditions; How was diagnosis reached?; Diagnostic biochemistry; Diagnostic biochemistry additional details; Diagnostic genetics; Diagnostic genetics additional details; Parental consanguinity; History of impaired fertility; History of a similar condition; Mother's height; Father's height; Research results .

*Visits:*

Complete DSD assessment at the ages closest to birth, 1 year, 5 years, 10 years and 15 years with the specific data requested for each age period.

*Additional information to be collected:*

- If performed brief description and date of partial gonadectomy
- Development of gonadal malignancies (even if diagnosed in different centres)

**Expected outputs**

Presentation - I-DSD Symposium, ESPE, National Meeting Italian Society of Andrology, National Congress Italian Society of Paediatric Endocrinology and Diabetology.

Publication.

**Publication Plan for authorship in outputs (refer to [guidance](#))**

All members of the study group and PIs of participating centres will be authors on all planned publications.

All authors will be required to meet all criteria listed below:

- Contributions to the conception, design, acquisition, analysis or interpretation of the work
- Drafting and revising the work
- Approving the final version of the work prior to dissemination
- Accountability for all aspects of the work