



Details of Study ID 202212_ALH

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<p>Date of Approval:</p> <p>December 2022</p>
<p>Name of study:</p> <p>Blood pressure in males with 46,XY DSD conditions</p>
<p>Summary of the proposed work to be performed with the registry data:</p> <p>Preliminary data from our studies show that people with hypospadias demonstrate some vascular features across the age span that may predispose them to cardiovascular disease and hypertension in adulthood. There is a need to pursue these studies further to understand whether this association is also present in any specific forms of DSD. This study will request blood pressure data on 46,XY DSD males registered on the I-DSD Registry. Data will then be compared between different forms of DSD to determine which groups, if any, are at increased risk of early hypertension. This will then allow the development of other studies in the future to assess cardiovascular risk.</p>
<p>Lay summary for the public (for the I-DSD/CAH websites and other publicity materials) (maximum 50 words)</p> <p>High blood pressure is a common cause of poor health in adults worldwide and can lead to heart disease. This study will seek to identify if boys or men with any particular DSD conditions have higher blood pressure readings by asking for any clinic or 24 hour blood pressure measurements over a certain time period. These readings will be compared between different groups to see if any have higher levels and may lead to the development of future studies looking at blood vessels in these groups.</p>
<p>Expected outputs</p> <p>Presentation – I-DSD Symposium, ESPE Publication</p>
<p>Publication Plan for authorship in outputs (refer to guidance)</p> <p>All members of the study group and PIs of participating centres will be authors on all planned publications.</p>



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

Inclusion criteria

46,XY

Raised as boys

Hypogonadism of non-adrenal origin e.g. disorder of androgen action/gonadal development

Consent to be included on the I-DSD Registry

Able to attend for BP and height measurements

Cases >2 years of age

Exclusion criteria

Other than 46,XY

Hypogonadism secondary to adrenal origin

Replacement with glucocorticoids, mineralocorticoids, or other steroid hormones

Raised as girls

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; Gestational age; Current gender; Karyotype (46,XY); Disorder type(all); Actual diagnosis (all); Associated conditions; Diagnostic genetics; Biobank sample available for research

Visits:

General: Date of assessment; Weight; Height; BMI

External phenotype: Meatus; Left gonad location; Right gonad location; Genital tubercle length; Labioscrotal fusion; Phallus size

Comorbidities: Hypertension

Medication: Testosterone; Other testosterone; DHT; Other DHT; Aromatase inhibitor; GnRH analogues; Glucocorticoids; Fludrocortisone; Oestrogen; Other Oestrogen; Other drugs; Other drugs note

Additional information (CRF collection):

Blood pressure at each visit