

Androgen Receptor, Implications for Health and Wellbeing: Natural History Study of Individuals With Androgen Insensitivity

NCT04708431

Status	RECRUITING
Sponsor	Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD)
Enrollment	650 participants

Key Eligibility Criteria

Inclusion (7)

- In order to be eligible to participate in this study, an individual must meet all the following criteria:
- Individuals ages 0-99 years old with known androgen insensitivity based on pathologic androgen receptor gene mutation or based on clinical diagnosis of complete androgen insensitivity (CAIS) based on 46 XY karyotype, presence of testis, absence of uterus, high testosterone without signs of virilization at birth or during puberty and/or multiple members in the family also presenting with clinical CAIS.
- Identify as male or female
- Patients with both complete, partial and mild androgen insensitivity are eligible
- Stated willingness to comply with all study procedures and availability for the duration of the study
- ... and 2 more (see full listing online)

Exclusion (3)

- An individual who meets any of the following criteria will be excluded from participation in this study: Patients with other diagnosis such as partial or complete gonadal dysgenesis, 5-alpha reductase deficiency, and 46 XY. If, following a diagnostic work-up, a patient is determined to have causes for 46 XY DSD other than androgen insensitivity; they will no longer be followed on this protocol. They will have the opportunity to continue care with the team under the Data Collection Protocol or may be referred to an expert or multidisciplinary DSD team in the community.
- Patients with significant non-endocrine medical conditions.
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Locations (1 total)

National Institutes of Health Clinical Center, Bethesda, Maryland, United States