Atypical Adult Presentations of Sexual Development in Individuals Presenting with Gender Dysphoria: Case Studies

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Disorders of Sexual Development

- congenital development of ambiguous genitalia (e.g., 46,XX virilizing congenital adrenal hyperplasia; clitoromegaly; micropenis)
- congenital disjunction of internal and external sex anatomy (e.g., Complete Androgen Insensitivity Syndrome; 5-alpha reductase deficiency)
- incomplete development of sex anatomy (e.g., vaginal agenesis; gonadal agenesis)
- sex chromosome anomalies (e.g., Turner Syndrome; Klinefelter Syndrome; sex chromosome mosaicism)
- disorders of gonadal development (e.g., ovotestes)

Association with gender variance

- Non-specific descriptions of association ie: GID-NOS
- Also an exclusion criteria for for GID in case definitions
- As a result the literature is most unhelpful.
- “The differences in phenomena, etiology, context of presentation, and treatment settings are so large that identical diagnoses and treatment approaches are not justified and may be detrimental…” (Meyer-Bahlburg H, 2009 IJT)

Clinical relevance of DSD in adults

- Child diagnoses: ***high phenotypic penetrance***, gender assignment is often the choice made by parents and clinicians (as late as possible to allow true gender to consolidate), the risk being iatrogenic GID
- Adult diagnoses: often present as gender dysphoria and are misdiagnosed as GID due to ***low phenotypic penetrance*** combined with limited clinical assessment
- Does it matter? Some patients want to know the 'why', health implications of some syndromes, sex designation changed with diagnosis, increased access to surgery

Clinical assessment tools

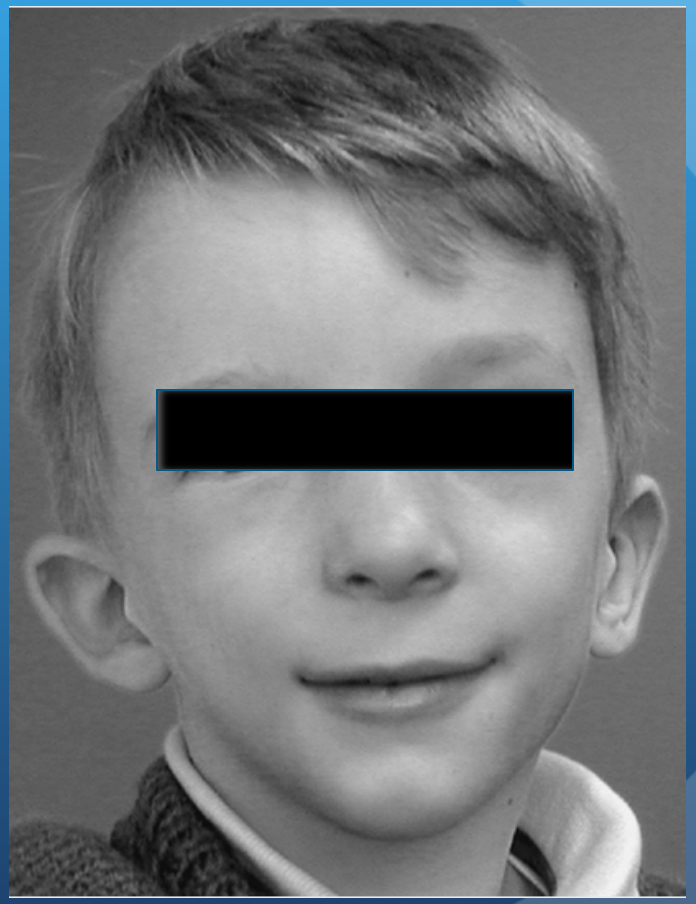
These should be standard:

- Prenatal history
- Childhood history
- Physical exam with tanner staging
- LH/FSH baseline

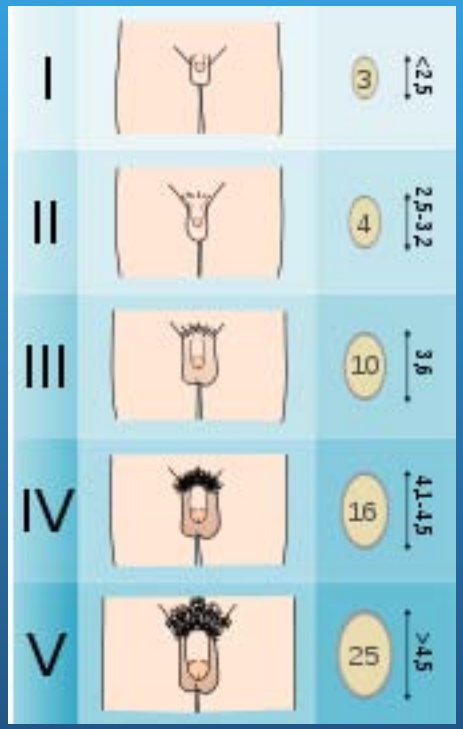
These are optional (as indicated) for investigation of DSD:

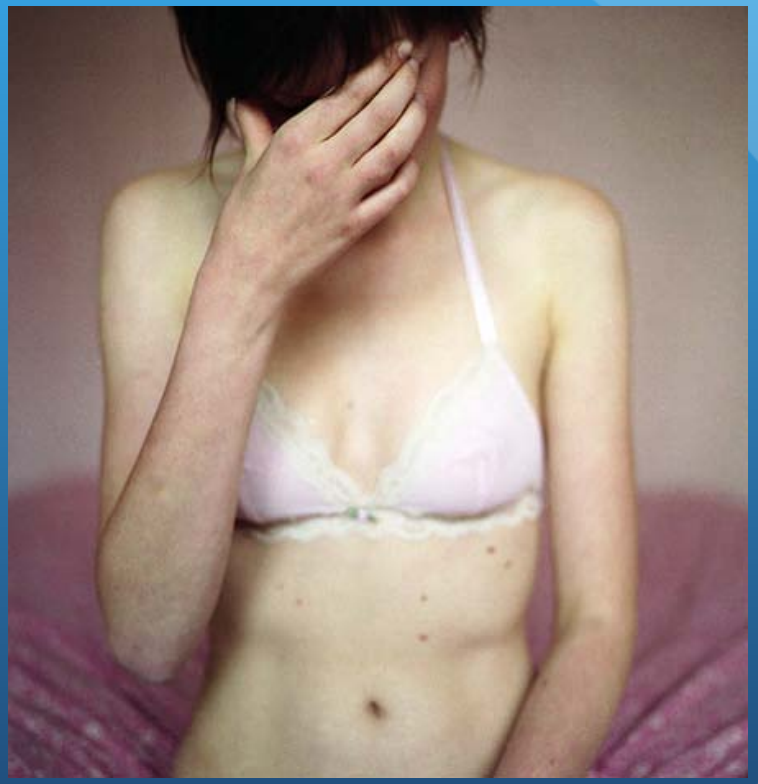
- Ultrasound imaging
- Karyotyping
- Other genetic/hormone/receptor tests

Case A: Russell Silver Syndrome

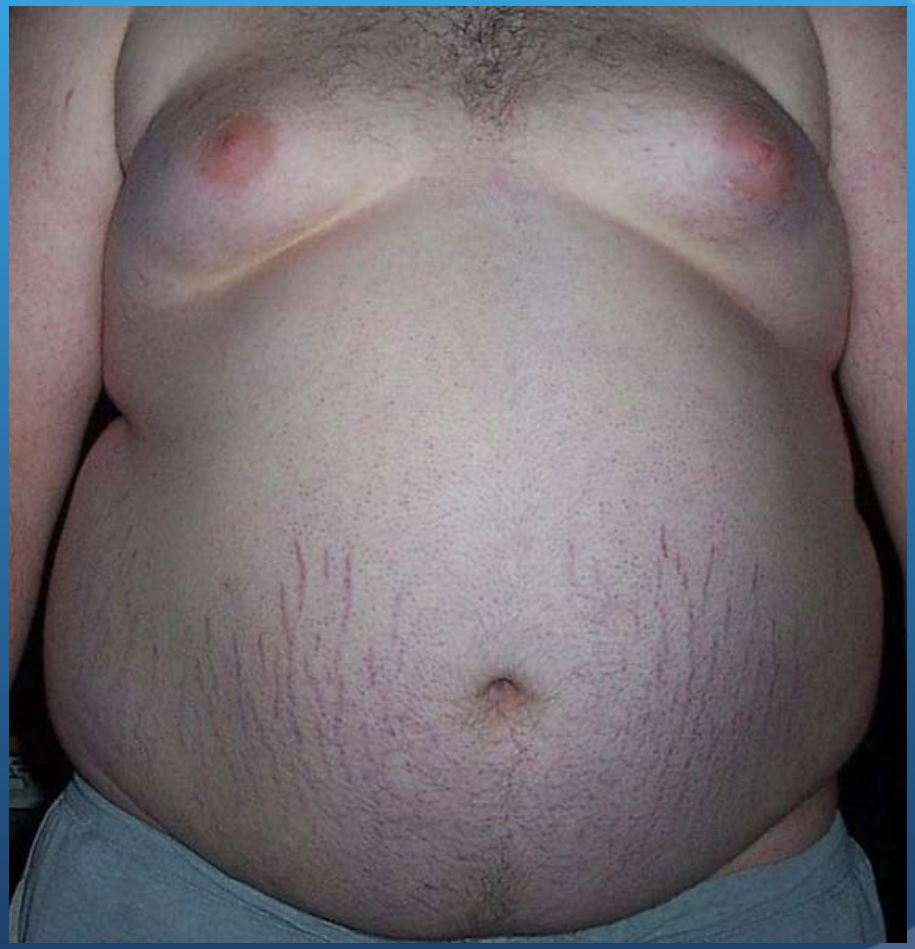


Case B: Androgen Insensitivity

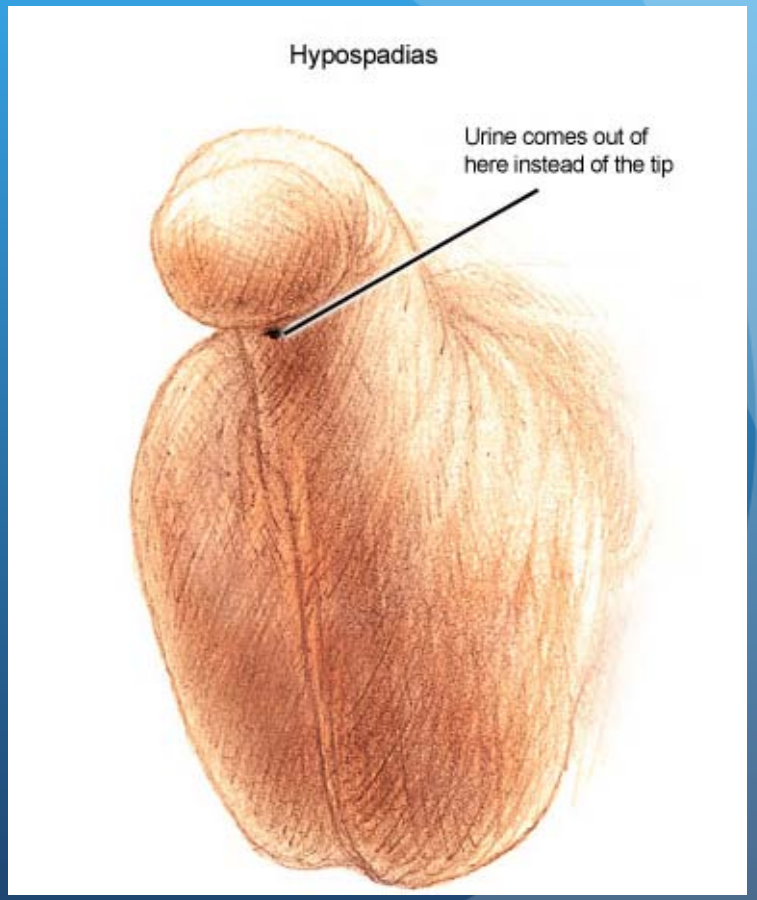




Case C: Polycystic ovarian syndrome



Case D: Ambiguous genetalia



Conclusion

- Persons with low penetrance phenotypic expression DSD often present with gender dysphoria in adulthood
- A thorough history and physical examination are essential if we are going to find them, and confirm gender congruence
- There are many reasons a person may want to know, but the most important is the access to treatment they are afforded