Clinical and Research-Focused Technical Review

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Background

Between December 2022 and June 2024, Equality Australia used Freedom of Information (FOI) legislation to obtain documents about medical procedures performed on intersex children in public hospitals across Australia that relate to their sex characteristics. Freedom of Information legislation requires organisations to release information unless there is an overriding public interest against their disclosure.

As a result of these requests, Equality Australia received 248 documents from children's hospitals across Australia. These documents contained information on 1444 total records of which:

- 1130 records provide insufficient detail for analysis. The majority were Medicare records from South Australia that contained no information on patient or rationale.
- 225 cases were too heavily redacted to sustain analysis.
- 9 cases were identifiably duplicate records.
- 64 unique cases capable of analysis - including 11 deferred procedures and 35 uncontroversial procedures.

17 cases that contained some cause for concern - inappropriate justifications for treatment (even if not determinative), the weight given to parental decision-making, or insufficient consideration of risks.

Equality Australia commissioned a technical review of the 83 cases for which analysis was feasible, with particular attention given to 17 cases previously identified as raising concerns. **I have reviewed these classifications and consider that the 17 cases were appropriately flagged.**

To illustrate the recurring themes and issues evident across this case set, I conducted a detailed evaluation of five cases selected from the FOI materials. This report presents my analysis of those five cases:

- Case 1 – Complex Hypospadias and Chordee
- Case 2 – 5-Alpha Reductase 2 Deficiency (5-ARD2)
- Case 3 – 46,XX CAH and Early Vaginoplasty
- Case 4 – 46,XX Presumed CAH: Gender Assignment and Timing of Surgery
- Case 5 – Turner Syndrome with Y-Chromosome Material (TS+Y)

Case analysis

Case 1: Complex Hypospadias and Chordee

FOI case number ref: FOI Document, *Doc ID 060* (NSW, 2020)

Date: October 2020

Age of patient: <1

Proposed surgery: Hypospadias

Rationale for surgery: Pros and cons of early surgery discussed with parents, parents want early surgery

MDT recommendation: Refer parents for psychological support, refer child for psychological counselling, further testing

Outcome: Early surgery likely

Infant with complex hypospadias and chordee; also hypopituitarism and biliary atresia. Biliary atresia is a condition requiring urgent early surgery to allow drainage of bile from the liver into the small intestine, and often necessitating liver transplantation later in life.

"Discussed that several operations may be necessary." The team plan was for hypospadias surgery and to consider DSD testing. Psychosocial support "could be considered if necessary."

The parents do not appear to have been given an in-depth exploration of proximal hypospadias surgery complications, which would be especially important in light of infant's medically-fragile status.

These complications are thoroughly summarized in a 2022 review[1] but were well known in 2020. The incidence of proximal hypospadias surgery complications can range from 15% to as high as 90%.[2]

This statistic is consistent even in centers performing many hypospadias procedures. For example, Children's Hospital of Philadelphia reported a 56% complication rate.[3] Each subsequent 'redo' urethroplasty has an even higher risk of complication.[4]

[1] Wood D, Wilcox D. Hypospadias: lessons learned. An overview of incidence, epidemiology, surgery, research, complications, and outcomes. Int J Impot Res. 2023 Feb;35(1):61-66. doi: 10.1038/s41443-022-00563-7. Epub 2022 Mar 29. PMID: 353520130.
[2] Alshafei A, Cascio S, Boland F, O'Shea N, Hickey A, Quinn F. Comparing the outcomes of tubularized incised plate urethroplasty and dorsal inlay graft urethroplasty in children with hypospadias: a systematic review and meta-analysis. J Pediatr Urol. 2020;16:154–61. https://doi.org/10.1016/j.jpurol.2020.01.009; Xiao D, Nie X, Wang W, Zhou J, Zhang M, Zhou Z, et al. Comparison of transverse island flap onlay and tubularized incised-plate urethroplasties for primary proximal hypospadias: a systematic review and meta-analysis. PLoS One. 2014;9: e106917. https://doi.org/10.1371/journal.pone.0106917; Saltzman AF, Carrasco A, Colvin A, Campbell JB, Vemulakonda VM, Wilcox D. Patients with disorders of sex development and proximal hypospadias are at high risk for reoperation. World J Urol. 2018;36:2051–8. https://doi.org/10.1007/ s00345-018-2350-3.
[3] Long CJ, Chu DI, Tenney RW, Morris AR, Weiss DA, Shukla AR, et al. Intermediate term followup of proximal hypospadias repair reveals high complication rate. J Urol. 2017;197:852–8. https://doi.org/10.1016/j.juro.2016.11.054.
[4] Wood DN, Andrich DE, Greenwell TJ, Mundy AR. Standing the test of time: the long-term results of urethroplasty. World J Urol. 2006;24:250–4. https://doi.org/ 10.1007/s00345-006-0057-3.

The most common complications are:

1. Fistula (abnormal opening between the urethra and the surface of the penis):
Fistula is the most common complication, occurring in up to 4–50% of patients. A fistula can occur any time, from acutely immediately after the urinary catheter is removed, to many years later.

2. Meatal stenosis (narrowing of the opening of the urethra):
Can present with difficulty urinating, a narrowed stream, spraying, or urinary tract infections secondary to incomplete bladder emptying.

3. Wound/glans dehiscence (opening of the surgical incision):
This frequently leads to urethral disruption that requires additional surgery.

4. Persistent chordee (curvature of the penis):
Persistent chordee is not usually detected early on but is increasingly seen in patients followed into puberty.

5. Diverticulae/urethroceles (sac-like outpouching of the urethral wall):
A urethrocele can present with a poor urinary stream, postvoid dribbling, urinary tract infections, penile swelling, and urethral stones.[5]

"It certainly seems that more proximal hypospadias is associated with a greater chance of long-term dissatisfaction with outcome."[6]

Although not published at the time of this case discussion, it is now known that risks of proximal hypospadias surgery are higher in the setting of a DSD diagnosis. According to a 2023 study by Scougall et al, those with a known diagnosis had a complication rate of 62%, and nearly half the complications arose 2 years after initial hypospadias repair.[7] This rate is higher than that of 56% previously reported in boys with proximal hypospadias.[8] Scougall et al state that "inherently, this cohort may be at a greater risk of complications due to the suboptimal anatomy associated with severe undermasculinization and DSD."[9]

[5] Wilcox D, Snodgrass W. Long-term outcome following hypospadias repair. World J Urol. 2006;24:240–3. https://doi.org/10.1007/s00345-006-0059-1; Keays MA, Dave S. Current hypospadias management: diagnosis, surgical management, and long-term patient-centred outcomes. Can Urol Assoc J. 2017;11: S48–53. https://doi.org/10.5489/cuaj.4386.
[6] Wood D, Wilcox D. Hypospadias: lessons learned. An overview of incidence, epidemiology, surgery, research, complications, and outcomes. Int J Impot Res. 2023 Feb;35(1):61-66. doi: 10.1038/s41443-022-00563-7. Epub 2022 Mar 29. PMID: 353520130.
[7] Scougall K, Bryce J, Baronio F, Boal RL, Castera JR, Castro S, Cheetham T, Costa EC, Darendeliler F, Davies JH, Dirlewanger M, Gazdagh G, Globa E, Guerra-Junior G, Guran T, Herrmann G, Holterhus PM, Akgül AK, Markosyan R, McElreavey K, Miranda ML, Nordenstrom A, O'Toole S, Poyrazoglu S, Russo G, Schwitzgebel V, Stancampiano M, Steigert M, Ahmed SF, Lucas-Herald AK. Predictors of surgical complications in boys with hypospadias: data from an international registry. World J Pediatr Surg. 2023 Oct 11;6(4):e000599. doi: 10.1136/wjps-2023-000599. PMID: 37860275; PMCID: PMC10582860.
[8] Long CJ, Chu DI, Tenney RW, Morris AR, Weiss DA, Shukla AR, et al. Intermediate term followup of proximal hypospadias repair reveals high complication rate. J Urol. 2017;197:852–8. https://doi.org/10.1016/j.juro.2016.11.054.
[9] Scougall K, Bryce J, Baronio F, Boal RL, Castera JR, Castro S, Cheetham T, Costa EC, Darendeliler F, Davies JH, Dirlewanger M, Gazdagh G, Globa E, Guerra-Junior G, Guran T, Herrmann G, Holterhus PM, Akgül AK, Markosyan R, McElreavey K, Miranda ML, Nordenstrom A, O'Toole S, Poyrazoglu S, Russo G, Schwitzgebel V, Stancampiano M, Steigert M, Ahmed SF, Lucas-Herald AK. Predictors of surgical complications in boys with hypospadias: data from an international registry. World J Pediatr Surg. 2023 Oct 11;6(4):e000599. doi: 10.1136/wjps-2023-000599. PMID: 37860275; PMCID: PMC10582860.

Case 2: 5-Alpha Reductase 2 Deficiency (5-ARD2)

FOI case number ref: FOI Document, *Doc ID 015* (NSW, 2018)**Date:** August 2018

Age of patient: <1

Proposed surgery: Hypospadias, Repair Orchidopexy

Rationale for surgery: Repair hypospadias, Parents want early surgery, Pros and cons of early surgery discussed with parents

MDT recommendation: Discuss surgery options with parents, Recommend early surgery

Outcome: Proceed with hypospadias at discretion of family / if family want to proceed with the surgery after discussion of potential risks

This family was originally seen in February 2018 (NSW - 2018.02 1802 – 2), when a presentation of "female genitalia with labial testes" was noted. The diagnosis was likely 5-alpha reductase 2 deficiency (5-AR2D), with genetic testing pending. At that time it was discussed that the literature supports raising these children as male because females "often" show preference for changing to male when virilization starts, but it was noted that outcomes for children who are "severely undervirilized" are difficult to predict. In a discussion of gender reassignment challenges, it was "not inappropriate for family to defer decision." The team recommended awaiting results of genetic testing before any gender reassignment or trying DHT cream (dihydrotestosterone) to see if penis enlarges, as well as ongoing close psychological support.

In August 2018, the time of this case discussion, 5-ARD2 is now genetically confirmed. The family changed gender to male. The team discussed orchiopexy, complex hypospadias repair, and recommended supporting surgery if the family wants it.

Why the rush to male assignment and surgery in a child with "female" genitalia? There is evidence that those assigned female with late-diagnosed 46,XY 5-ARD2 and mild genital difference can identify as female in adulthood.[10] Genital surgery risks reinforcing misassigned gender.

Case 3: 46,XX CAH and Early Vaginoplasty

FOI case number ref: FOI Document, *Doc ID 022* (NSW, 2018)

Date: November 2018

Age of patient: <1

Proposed surgery: Vaginoplasty

[10] Nascimento RLP, de Andrade Mesquita IM, Gondim R, Dos Apóstolos RAAC, Toralles MB, de Oliveira LB, Canguçu-Campinho AK, Barroso U Jr. Gender identity in patients with 5-alpha reductase deficiency raised as females. J Pediatr Urol. 2018 Oct;14(5):419.e1-419.e6. doi: 10.1016/j.jpurol.2018.08.021. Epub 2018 Sep 5. PMID: 30297225; Berra M, Williams EL, Muroni B, Creighton SM, Honour JW, Rumsby G, Conway GS. Recognition of 5αreductase-2 deficiency in an adult female 46XY DSD clinic. Eur J Endocrinol. 2011 Jun;164(6):1019-25. doi: 10.1530/EJE-10-0930. Epub 2011 Mar 14. PMID: 21402750.

Rationale for surgery: Child unlikely to remember surgery, better healing, urinary tract infection prevention, parents want early surgery

MDT recommendation: Recommend early surgery

Outcome: Vaginoplasty for infant with CAH. Reasons included "avoiding stigmatisation of genital variation by restoring female anatomy, preventing parental anxiety"

Infant with 46,XX CAH and Prader 3 genital difference (enlarged clitoris, near-complete fusion of labioscrotal folds, vagina and urethra open internally into a common urogenital sinus with a single opening on the outside). Parents want early vaginoplasty.

"The group felt that the requested surgery was I keeping with current guidelines and the family should be clearly be made aware of the risks (may need revision at puberty, anaesthetic risks) and benefits (lack of memory of procedure, better healing, UTI [urinary tract infection] prevention, avoiding stigmatization of genital variation by restoring female anatomy, preventing parental anxiety) of early surgery versus benefits of late surgery (patient involved in decision making less risk of revision surgery). The family should be made clearly aware that *There is no objective evidence at this time as to whether early, late, or no surgery best preserves overall QOL or sexual function.*

Recommendations:

1. The group unanimously agreed to support the parents and treating clinicians' request to proceed with vaginoplasty as per international guidelines as long as the pros and cons of early versus late surgery were clearly discussed with the family.
2. Clitoroplasty has not been requested and could be delayed until the child is older."

The team cites Endocrine Society Guidelines,[11] as well as reasons unsubstantiated by evidence to support early surgery.

There is no evidence for "better healing" in early surgery because there are no studies comparing healing in early vs. late surgery. Since there is no research directly comparing outcomes of early and late feminizing genitoplasty (FG), we cannot know which one has better healing, although some gynecologists who perform both primary FG as well as surgery to treat subsequent complications in older patients advocate for deferral.[12]

It was commonly believed in the past that early FG prevents UTIs, as implied in 2002 Pediatric Endocrine Society guidelines.[13] Today we know that girls with CAH who have a common urogenital sinus are not predisposed to UTI prior to surgery, and an intact urogenital sinus does not predispose to UTI later.[14]

In spite of these facts, many families continue to testify anecdotally that they have been told FG will prevent UTI. Surgery also does not prevent significant non-infectious urinary issues. In long-term

[11] Speiser PW, Azziz R, Baskin LS, Ghizzoni L, Hensle TW, Merke DP, Meyer-Bahlburg HF, Miller WL, Montori VM, Oberfield SE, Ritzen M, White PC; Endocrine Society. Congenital adrenal hyperplasia due to steroid 21-hydroxylase deficiency: an Endocrine Society clinical practice guideline. J Clin Endocrinol Metab. 2010 Sep;95(9):4133-60. doi: 10.1210/jc.2009-2631. Erratum in: J Clin Endocrinol Metab. 2010 Nov;95(11):5137. Erratum in: J Clin Endocrinol Metab. 2021 Jun 16;106(7):e2853. doi: 10.1210/clinem/dgab316. PMID: 20823466; PMCID: PMC2936060.
[12] Creighton, S., et al., Timing and nature of reconstructive surgery for disorders of sex development - introduction. J Pediatr Urol, 2012. 8(6): p. 602-10; Murphy, C., L. Allen, and M.A. Jamieson, Ambiguous genitalia in the newborn: an overview and teaching tool. J Pediatr Adolesc Gynecol, 2011. 24(5): p. 236-50.
[13] Consensus statement on 21-hydroxylase deficiency from the Lawson Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology. J Clin Endocrinol Metab, 2002. 87(9): p. 4048-53.
[14] Nabhan, Z.M., R.C. Rink, and E.A. Eugster, Urinary tract infections in children with congenital adrenal hyperplasia. J Ped Endocrinol Metab, 2006. 19: p. 815-820.

follow up of adults, whether they had surgery or not, adults with CAH were more likely to have urinary symptoms, particularly incontinence, than age-matched controls, and those with urinary symptoms were 9 times as likely as symptomatic controls to report an adverse effect on their lives.[15] Urinary outcomes in adults are poorly- studied.[16]

Stigma in adults with CAH, which surgery is meant to prevent, is perennially cited as an issue of concern to be prevented by "normalizing" surgery.[17]

Several studies documented that stigma was experienced by adult women with CAH in a variety of settings, even though most of the women had previous surgery. Stigma experienced by nearly 2/3 of adults in the general social environment was related to obvious physical differences, such as hirsutism or a deep voice, rather than genital difference.[18]

25% of the same women reported that doctors' actions caused stigma, mostly via frequent genital exams in teaching settings.[19] This is a significant finding because despite years of patient complaints about traumatizing genital exams, the practice continues in contemporary multidisciplinary clinics, of which the US National Institutes of Health-funded Translational Research Network (TRN) found that 30% still perform genital exams for teaching.[20]

Sexual stigma was experienced by 40% of the women studied, whether they had surgery (the majority) or not, but nearly all women described maladaptive coping in interviews, including secrecy, hiding genitalia, sex avoidance or abstinence, and substance abuse.[21]

Rather than being a consequence of genital difference, shame can result from the mere fact of having genitals that "required surgery," suggesting significant iatrogenic moderators of the relationship between genital difference and sexual stigma.[22]

Case 4 - 46,XX Presumed CAH: Gender Assignment and Timing of Surgery

FOI case number ref: FOI Document, *Doc ID 131* (QLD, 2022)

Date: 13 September 2013

Age of patient: <1

Proposed surgery: Vaginoplasty

Rationale for surgery: Non-functioning, Parents want early surgery, UTI prevention

[15] Davies, M.C., et al., Congenital adrenal hyperplasia and lower urinary tract symptoms. BJU Int, 2005. 95(9): p. 1263-6.
[16] Cools, M., et al., Caring for individuals with a difference of sex development (DSD): a Consensus Statement. Nat Rev Endocrinol, 2018. 14(7): p. 415-429.
[17] Mouriquand, P.D., et al., Surgery in disorders of sex development (DSD) with a gender issue: If (why), when, and how? J Pediatr Urol, 2016. 12(3): p. 139-49.
[18] Meyer-Bahlburg, H.F., et al., Syndrome-Related Stigma in the General Social Environment as Reported by Women with Classical Congenital Adrenal Hyperplasia. Arch Sex Behav, 2017. 46(2): p. 341-351.
[19] Meyer-Bahlburg, H.F., et al., Stigma in Medical Settings As Reported Retrospectively by Women With Congenital Adrenal Hyperplasia (CAH) for Their Childhood and Adolescence. J Pediatr Psychol, 2016. 2015: p. 1-8.
[20] Rolston, A.M., et al., Disorders of sex development (DSD): Clinical service delivery in the United States. Am J Med Genet C Semin Med Genet, 2017. 175(2): p. 268-278.
[21] Meyer-Bahlburg, H.F.L., et al., Stigma Associated with Classical Congenital Adrenal Hyperplasia in Women's Sexual Lives. Arch Sex Behav, 2017: p. DOI 10.1007/s10508-017-1003-8.
[22] Meyer-Bahlburg, H.F.L., et al., Stigma Associated with Classical Congenital Adrenal Hyperplasia in Women's Sexual Lives. Arch Sex Behav, 2017: p. DOI 10.1007/s10508-017-1003-8.

MDT recommendation: Defer surgery decision to later date, Discuss surgery options with parents, Further testing

Outcome: Strong parental distress noted around not having early surgery including vaginoplasty, and clinicians' notes on changes in surgery practices in Australia. Will need surgery prior to puberty given no separate vaginal opening. Two surgical consultants discussed timing of vaginoplasty and could take place in next 12 months for urogenital compromise given high risk of infection.

46,XX presumed CAH ; gene testing pending. "Virilised: phallus >2cm, urethral meatus on ventral surface, no vaginal opening, labioscrotal folds fused."

The discussion noted "controversy regarding timing of surgery." The "majority of people" will have female gender identity, but no statistics are quoted.

The parents are worried about recurrent UTIs, and feel that trauma would be "minimized" with early surgery. They question if the team is using "evidence-based decision-making frameworks, and whether decision may be unduly influenced by fear of potential future litigation."

The team is concerned that the parents are distressed about delaying surgery until the child can consent, and that human rights frameworks centering children's autonomy give inadequate consideration to parental rights.

The group discussed variations in practice around Australia regarding early surgery in CAH. Referring to "radical clitorectomy" in past as the source of sexual difficulties, current "practice within QLD has been for vaginoplasty… with some clitoral *recession* [italics original] rather than resection. This allows for preservation of sensation and clitoral function."

The team planned further imaging and discussion.

Realistic presentation of what is known about sexual function and sensation after clitoral surgery did not occur, such as evidence from a 2018 literature review by Almasri et al.:

> "Females with congenital adrenal hyperplasia (CAH) and atypical genitalia often undergo complex surgeries; however, their outcomes remain largely uncertain. Methods: We searched several databases through 8 March 2016 for studies evaluating genital reconstructive surgery in females with CAH. Reviewers working independently and in duplicate selected and appraised the studies. Results: We included 29 observational studies (1178 patients, mean age at surgery, 2.7 +/- 4.7 years; mostly classic CAH). After an average follow-up of 10.3 years, most patients who had undergone surgery had a female gender identity (88.7%) and were heterosexual (76.2%). Females who underwent surgery reported a sexual function score of 25.13 using the Female Sexual Function Index (maximum score, 36). Many patients continued to complain of substantial impairment of sensitivity in the clitoris, vaginal penetration difficulties, and low intercourse frequency. Most patients were sexually active, although only 48% reported comfortable intercourse. Most patients (79.4%) and treating health care professionals (71.8%) were satisfied with the surgical outcomes. Vaginal stenosis was common (27%), and other surgical complications, such as fistulas, urinary incontinence, and urinary tract infections, were less common. Data on quality of life were sparse and inconclusive. Conclusion: The long-term follow-up of females with CAH who had undergone urogenital reconstructive surgery shows variable sexual function. Most patients were sexually active and satisfied with the surgical outcomes; however, some patients still complained of

impairment in sexual experience and satisfaction. The certainty in the available evidence is very low."[23]

A more complete picture of the evidence regarding female gender identity was also not presented. Publications favoring early surgery in CAH contain statements such as, "female assignment is suggested for those with 46,XX and CAH, since 95% develop female gender identity," or "there is usually no gender issue in this group," but the literature in this area has serious faults.[24]

In children with CAH, as in all children, gender identity is a result of "complex, multiple and interactive developmental processes."[25] It is not fixed at birth, nor is it confirmed by "fixing" genitals with surgery aimed at creating dimorphism.

Many studies of gender outcomes in CAH using binary outcomes have methodologic limitations. Early surgery proponents minimize the significance of surgical reinforcement of gender misassignment with claims that multiple studies show a low rate of gender dysphoria in CAH. A 2015 literature review by Pasterski et al found the results of older studies often cited as supporting early surgical reinforcement of female gender assignment to be unreliable because they used flawed methodologies including inconsistent, insufficient, or unvalidated measurements; even those using measurements based on American Psychiatric Association Diagnostic and Statistical Manual (DSM) or self-report questionnaires/interviews confounded gender identity with gender role behaviors.[26] While older literature on gender in DSD conflated diverse gender identity and expression with gender dysphoria (GD), current DSM-5 criteria distinguish GD from gender diversity by including a strong desire to be a different gender.[27]

Among those studies discredited in analysis by Pasterski et al is one that is frequently cited by surgery proponents for its numerical significance, a 2005 literature analysis by Dessens et al that reports on 250 people.[28]

Following the work of Pasterski et al, a large international study was published in 2018. In "Gender Dysphoria and Gender Change in Disorders of Sex Development/Intersex Conditions: Results from the dsd-LIFE Study," which is the largest study investigating gender outcomes in intersex/DSD, the data collected ostensibly show a 0.4% rate of GD in CAH, but the authors themselves caution that multiple methodologic issues challenge the study's validity.[29] Although it was a mixed methods study, quantitative questionnaires were not developed on the basis of clinical interviews. Of 221 female assigned participants with CAH, 174 had confirmed CAH, but 47 were not specified. "Because their gender did not correspond with the usual gender for their diagnosis," those living as male were excluded. Finally, 36% of scores on questions meant to assess GD were missing; only questions on recent sexual activity had a similar rate of missing responses. Lack of response to questions may reflect wording that unintentionally stigmatizes or pathologizes gender diversity.

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[23] Almasri J, Zaiem F, Rodriguez-Gutierrez R, Tamhane SU, Iqbal AM, Prokop LJ, Speiser PW, Baskin LS, Bancos I, Murad MH. Genital Reconstructive Surgery in Females With Congenital Adrenal Hyperplasia: A Systematic Review and Meta-Analysis. J Clin Endocrinol Metab. 2018 Nov 1;103(11):4089-4096. doi: 10.1210/jc.2018-01863. PMID: 30272250.
[24] Mouriquand, P.D., et al., Surgery in disorders of sex development (DSD) with a gender issue: If (why), when, and how? J Pediatr Urol, 2016. **12**(3): p. 139-49.
[25] Liao, L.M., et al., Determinant factors of gender identity: a commentary. J Pediatr Urol, 2012.**8**(6): p. 597-601.
[26] Pasterski, V., et al., Increased Cross-Gender Identification Independent of Gender Role Behavior in Girls with Congenital Adrenal Hyperplasia: Results from a Standardized Assessment of 4- to 11-YearOld Children. Arch Sex Behav, 2015. **44**(5): p. 1363-75.
[27] American Psychiatric Association. Diagnostic and statistical manual of mental disorders: DSM-5. 2013: American Psychiatric Association.
[28] Dessens, A.B., F.M. Slijper, and S.L. Drop, Gender dysphoria and gender change in chromosomal females with congenital adrenal hyperplasia. Arch Sex Behav, 2005. **34**(4): p. 389-97.
[29] Kreukels, B.P.C., et al., Gender Dysphoria and Gender Change in Disorders of Sex Development/Intersex Conditions: Results From the dsd-LIFE Study. J Sex Med, 2018. **15**(5): p. 777785.

In order to avoid methodologic limitations, Pasterski et al performed their own study *prospectively* assessing gender identity of 81 female-assigned 4- to 11year old children with CAH using mixed qualitative and quantitative methods, including the gold standard, DSM-5 Part A criteria for gender dysphoria.[30] They found that cross-gender identification was significantly increased in these children relative to both XY siblings with CAH and unaffected siblings. The results in 12% of female-assigned children met all 5 DSM Part A criteria for GD, which would qualify them for referral to a GD clinic. 12% is hardly rare; it equates to 1 out of 8 patients, the same as the proportion of women who will develop breast cancer in their lifetimes, which is not considered at all unusual. It is also nearly 20 times higher than the estimated rate of GD in non- intersex children (0.7%).[31]

Among all studies of adult gender identity outcomes, there is one that stands out for utilizing the type of mixed methods- interviews plus quantitative scales developed from those interviews- recommended by the International Rare Diseases Research Consortium (IRDiRC) for research on rare conditions.[32]

Schweizer et al studied 69 people with diverse intersex/DSD, including 17 patients with CAH.[33] Although the sample size is small, the investigation yielded details unmatched in richness, providing complex and nuanced insights not found in other studies. Among those 17 patients, with one nonresponder, 11/16 (69%) identified as women, 4/16 (25%) reported a 'mixed' two-gender identity, and 1/16 (6%) a male gender identity. 10 of 12 of those originally assigned female (2 born with female genitalia and 8 with ambiguity), were fairly to highly satisfied with assignment (83%). Among the satisfied 10, however, 1 had mixed identity. Of the 2 (17%) not satisfied with female birth assignment, both had genital ambiguity; 1 was reassigned male at age 7 based on medical recommendations following signs of male development, and continued living as male but had a mixed 2 gender identity. The other person not satisfied with female assignment had mixed identity and lived in a 3rd gender in adulthood. 5 people were assigned male before age 1, 4 having male genitalia; 1 (20%) was ultimately satisfied and lived in a male role. Two who were reassigned female before age 2 (1 with ambiguous and 1 with male genitalia at birth) later identified and lived as female. One person with male genitalia at birth and assigned male, who was reassigned female before age 1 and self-reassigned male at 35, had mixed gender identity and lived in a 3rd gender. The 5th person assigned male, who had male genitalia at birth and underwent many medical male-sex-assigning interventions, later identified as female and was considering a male to female gender transition.

The results show that gender assignment based on genital appearance alone is not predictive of adult gender identity. There is significant dissatisfaction with gender assignment, both male and female, even in the absence of gender transition. Among the study's surprising findings was that 7/16 (41%) people, including some who were content with gender assignment, had markedly low scores on the certainty of belonging to one specific gender (CG) scale. These people might be described using American Academy of Pediatrics language as gender diverse.[34]

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[30] Pasterski, V., et al., Increased Cross-Gender Identification Independent of Gender Role Behavior in Girls with Congenital Adrenal Hyperplasia: Results from a Standardized Assessment of 4- to 11-YearOld Children. Arch Sex Behav, 2015. **44**(5): p. 1363-75.
[31] Herman, J.L., et al. Age of individuals who identify as transgender in the United States. 2017; Williams Institute UCLA. Available from: (http://williamsinstitute.law.ucla.edu/wp- content/uploads/TransAgeReport.pdf).
[32] Morel, T. and S.J. Cano, Measuring what matters to rare disease patients - reflections on the work by the IRDiRC taskforce on patient-centered outcome measures. Orphanet J Rare Dis, 2017. **12**(1): p. 171.
[33] Schweizer, K., et al., Gender experience and satisfaction with gender allocation in adults with diverse intersex conditions (divergences of sex development, DSD). Psychology & Sexuality, 2013. **5**(1): p. 56-82.
[34] Rafferty, J., AAP Policy Statement. Ensuring Comprehensive Care and Support for Transgender and Gender-Diverse Children and Adolescents. Pediatrics, 2018. **142**(4).

Schweizer et al concluded that their findings indicate

> "… the inadequacy of the dichotomous, one-dimensional male/ female categorization for the purpose of allowing an authentic sense of gender identity in individuals with DSD. Our research further suggests that treatment goals should be re-directed from 'successful' gender outcome in binary terms to psychological well-being regardless of feeling male, female, both or neither."[35]

The finding that 25% of people with CAH have identities not subsumed in current terminology makes it clear that more expansive understandings of gender as dynamic and non-binary are needed.

With errors in early childhood gender assignment a significant possibility, deferring surgery in children preserves options for later transition. Social assignment is easily changed, but irreversible surgery compounds the magnitude of harm from misassignment to catastrophic proportions, as in the removal of a healthy penis from a child subsequently identifying as male. As told to the researchers, "The definition as female and the iatrogenic trauma connected with it destroys identity."[36]

Greater gender diversity in DSD/intersex than in the general population is expected, given genetic factors, varying hormonal exposure, and the psychological/emotional effects of living with variant sex traits.[37] A gender affirmative model of care for intersex/DSD should address gender contentedness, discomfort or distress, and avoid stigmatizing diverse identities and expression. Findings of true gender dysphoria should prompt appropriate support and intervention.

Case 5: Turner Syndrome with Y-Chromosome Material (TS+Y)

FOI case number ref: FOI Document, *Doc ID 056* (NSW, 2020)

Date: August 2020

Age of patient: 8-10

Proposed surgery: Gonadectomy

Rationale for surgery: Risk of malignancy, Non-functioning

MDT recommendation: Recommend early surgery, Hormone treatment

Outcome: Risk of gonadoblastoma redacted

Phenotypically female child referred to endocrinologist for growth hormone (GH) treatment. Prenatal diagnosis of probable mosaic Turner syndrome based on clinical features. She has dysgenetic internal gonads with Y material.

[35] Schweizer, K., et al., Gender experience and satisfaction with gender allocation in adults with diverse intersex conditions (divergences of sex development, DSD). Psychology & Sexuality, 2013. **5**(1): p. 56-82.
[36] Schweizer, K., et al., Gender experience and satisfaction with gender allocation in adults with diverse intersex conditions (divergences of sex development, DSD). Psychology & Sexuality, 2013. **5**(1): p. 56-82.
[37] Cools, M., et al., Caring for individuals with a difference of sex development (DSD): a Consensus Statement. Nat Rev Endocrinol, 2018. **14**(7): p. 415-429.

"Although there is no proof that growth hormone treatment would accelerate a malignancy, clinical practice has been for gonadectomy prior to GH treatment in view of the high risk of atypical features in dysgenetic intra-abdominal gonads with Y material…The group recommended bilateral gonadectomy prior to commencing growth hormone treatment but irrespective of whether growth hormone treatment occurs, consistent with current international guidelines."

Regarding the timing of gonadectomy in mosaic Turner Syndrome with Y material (TS+Y), a 2020 study looked at rates of spontaneous thelarche (breast development) and menarche in pubertal-age children so that factors related to ovarian function and fertility potential could be incorporated into decision-making.[38] It found a 42% chance of spontaneous thelarche and 11% chance of spontaneous menarche, with a 3% rate of gonadal malignancy. There have been case reports of pregnancy in women with TS+Y; spontaneous thelarche and menarche are associated with fertility in TS.[39] A 2025 study confirmed a 3.5% risk of malignant transformation, and that prior exposure to growth hormone was not predictive of development of gonadal tumors.[40] Infertility is an important consideration for patients and families, and fertility preservation should be part of discussions of gonadectomy timing in children with TS+Y.

[38] Dabrowski E, Johnson EK, Patel V, Hsu Y, Davis S, Goetsch AL, Habiby R, Brickman WJ, Finlayson C. Turner Syndrome with Y Chromosome: Spontaneous Thelarche, Menarche, and Risk of Malignancy. J Pediatr Adolesc Gynecol. 2020 Feb;33(1):10-14. doi: 10.1016/j.jpag.2019.08.011. Epub 2019 Aug 26. PMID: 31465855; PMCID: PMC7413626.

[39] Borgstrom B, Hreinsson J, Rasmussen C, et al: Fertility preservation in girls with Turner syndrome: prognostic signs of the presence of ovarian follicles. J Clin Endocrinol Metab 2009; 94:74.

[40] Dowlut-McElroy T, Long JR, Mayhew AC, Lawson A, Fei YF, Smith AK, Shankar RK, Gomez-Lobo V. Gonadal Tumors in Individuals with Turner Syndrome and Y-Chromosome Mosaicism: A Retrospective Multisite Study. J Pediatr Adolesc Gynecol. 2025 Apr;38(2):154-160. doi: 10.1016/j.jpag.2024.11.005. Epub 2024 Nov 20. PMID: 39577758.