Hormones, context, and “Brain Gender”: A review of evidence from congenital adrenal hyperplasia

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**Abstract**

Brain organization theory suggests that steroid hormones during fetal development permanently organize the brain for gender, including patterns of sexuality, cognition, temperament, and interests that differ by sex. This widely-accepted theory has important implications for health, ranging from medical management of infants with intersex conditions to suggested etiologies for sex differences in autism, depression, and other mental health problems. Studies of genetic females with congenital adrenal hyperplasia (CAH), in which high prenatal androgens have been linked to both atypical genitals and “masculine” patterns of gender and sexuality, are particularly important. Based on a comprehensive review of research on CAH, this article demonstrates that such studies have neglected four broad categories of variables that plausibly affect psychosexual development: 1) physiological effects of CAH, including complex disruption of steroid hormones from early development onwards; 2) intensive medical intervention and surveillance, which many women with CAH describe as traumatic; 3) direct effects of genital morphology on sexuality (versus indirect effects that “masculine” genitals may have on gender socialization); and 4) expectations of masculinization that likely affect both the development and evaluation of gender and sexuality in CAH. Complex and iterative interactions among postnatal biological variables, medical interventions, and social context provide a more plausible explanation for atypicalities in psychology and behavior that have been reported for genetic females with CAH than the conventional explanation that early androgens have “masculinized” their brains.

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**Introduction**

Brain Organization Theory suggests that steroid hormones during fetal development permanently organize the brain for gender, including patterns of sexuality, cognition, temperament, interests, and mental health that are considered “masculine” or “feminine”. Widely accepted in both popular and scientific literature (e.g., Hines, 2004; Institute of Medicine, 2001, 121; Pinker, 2005), the theory has important implications for health, from medical management of infants with intersex conditions to prevention and treatment of mental health problems for which there are male-female differences in prevalence, such as anxiety, autism, and depression (Bailey & Hurd, 2005; Knickmeyer et al., 2006). Obviously, the theory that prenatal hormones in utero organize the brain for gender-typical traits cannot be experimentally investigated in humans, and the only comprehensive review of human studies found that evidence across the various observational research designs does not agree, as it must for the theory to be supported as a general explanation for human cognitive and psychosexual development (Jordan-Young 2010). That same review, however, found potential support for the theory from studies of congenital adrenal hyperplasia (CAH). In classical CAH, androgen levels are high during fetal development, causing genetic females to be born with atypical or male-appearing genitalia (White & Speiser, 2000). There is also broadly consistent evidence that girls and women with CAH differ from comparison groups on traits that include gender-typical toy preferences (e.g., Berenbaum & Hines, 1992; Pasteriks et al., 2005; Servin, Nordenstrom, Larsson, & Bohlin, 2003) and sexual orientation (Meyer-Bahlburg & Dolezal, 2008; Zucker et al., 1996). The nearly universally-accepted explanation for atypical psychosexuality in CAH is that elevated androgens have masculinized the brain early in development.

Studies of CAH are therefore extremely important for brain organization theory. Since human brain organization studies, taken as a whole, do not support the theory, the apparent compatibility of CAH studies with hormonal brain organization begs for exploration. Both critics and proponents of the theory highlight one alternative explanation for the observed differences in
psychosexuality between CAH-affected and unaffected females: the indirect effects of masculinized genitalia on early gender socialization (Bleier, 1984; Longino & Doell, 1983). Atypical genitalia surely affect developmental experiences in important ways that include parental anxiety or uncertainty about the child's sex, and there is evidence that early treatment does not eliminate these concerns (Karkazis, 2008). In fact, as I shall argue below, early, usually multiple, surgeries and treatments such as vaginal dilation may amplify, rather than eliminate, the consequences of atypical genitalia, though proponents of brain organization theory ignore this possibility (e.g., Hines, 2010). This is just one of the many factors of postnatal development, including physiological effects as well as intensive medical monitoring, which plausibly affect psychosexuality in girls and women with CAH. Yet brain organization studies narrowly focus on early hormone exposures, to the exclusion of virtually all other variables that affect development.

In this article, I explore four general categories of variables that brain organization studies omit, but which likely affect psychosexual development and behavior. First, physiological effects of CAH include profound effects on metabolism, neuroendocrine function, and gross (non-genital) morphology. Second, intensive medical management and surveillance, typically from shortly after birth in classical CAH, has been assumed to minimize, rather than produce, psychosocial patterns associated with CAH. Third, masculinized genitals are sometimes discussed in brain organization studies, but discussions are limited to the question of whether masculine genitals cause parents to be confused or ambivalent about gender, and therefore encourage masculine behavior in girls with CAH. Remarkably, the more obvious direct effects of atypical genitals on sexuality are ignored. Finally, expectations of masculinization primed by brain organization theory itself likely affect both the development of gender and sexuality, and the way that traits and behaviors are perceived and reported.

Methodology

For this article, I sought to identify all brain organization research on genetic females with CAH, defined as studies that report on aspects of psychosocial behavioral gender or sexuality in this group, published from 1967 (when brain organization theory was first applied to humans) through mid-2010. I searched the ISI Web of Science database for original, peer-reviewed research reports on gender or sexuality in genetic females with CAH, limiting to English language articles, 1967–2010. I combined terms for “congenital adrenal hyperplasia” (including earlier nomenclature for the syndrome, such as “adrenogenital syndrome” and “congenital virilizing hyperplasia”) with terms for behavior, cognition, gender, or psychosexuality (such as masculinity, femininity, eroticism, sexual behavior, sexual orientation). The search yielded 423 articles; of these, I retained 103 articles for analysis after scanning titles and abstracts (e.g., to exclude review articles, articles that mentioned CAH but did not in fact present new psychosexual or behavioral data on CAH, and commentary/opinion pieces). I also reviewed medical, psychological, and anthropological literature that described clinical features in and experiences of people with CAH, in order to identify and explore a wider range of biological and social context factors that may contribute to “gender-atypical” behavior associated with CAH. I identified four categories of omitted variables (non-genital physiological effects of CAH, intensive medical intervention and surveillance, the direct effects of atypical genitalia on sexuality, and labeling and priming effects). Following the method of feminist theoretical and empirical work on gender and embodiment (e.g., Bordo, 1993; Fausto-Sterling, 2005; Toerien, Wilkinson, & Choi, 2005), I explored how specific physical manifestations of CAH as well as responses to CAH may become entangled with the lived experience of gender for girls and women with this condition.

Clinical and behavioral features in CAH

CAH is a genetically-transmitted condition that impairs adrenal synthesis of cortisol, a steroid hormone that is important for a wide range of metabolic activities, tissue development, and immune function. CAH affects both males and females, occurring in both a “classical” early form (affecting roughly 1 in 16,000 live births in most populations) and a milder, late-onset form (affecting 0.2–2% of live births, depending on population) (Speiser & White, 2003). Most cases of CAH are caused by a deficiency in the 21-hydroxylase enzyme (Nimkarn & New, 2009), which interferes with cortisol production and increases androgen production beginning around the same time as the genitals are undergoing sex-differentiation. In genetic females, high levels of prenatal testosterone and duffy-protestosterone in the classical form cause various degrees of genital masculinization, from a larger-than-usual clitoris to a fully-formed penis; labia can be fully fused, resembling an empty scrotum (White & Speiser, 2000); nearly a quarter of genetic females with classical CAH are initially assigned as males (Dessens, Slipper, & Drop, 2005). About 75% of people with classical CAH have the severe “salt-wasting” form (Lekarev, Parsa, Nimkarn, Lin-Su, & New, 2010) which is also associated with more extreme genital anomalies. Elevated steroids cause progressive postnatal problems, such as premature physical maturation and puberty, in both sexes. This article focuses on CAH in genetic females, because claims (and concerns) about brain masculinization are built on these cases.

There is evidence of behavioral and psychological differences between genetic females with and without CAH (Cohen-Bendahan, van de Beek, & Berenbaum, 2005; Hines, 2004), though the specific behaviors affected, the degree of difference, and the extent to which CAH-associated traits are properly considered “masculine” is often overstated. There have been reports of elevated “male-typical” traits such as assertiveness and aggression, rough and tumble play, IQ, specific cognitive abilities, and “masculine” sexuality in girls and women with CAH (e.g., Berenbaum & Hines, 1992; Berenbaum & Resnick, 1997; Erhardt, Epstein et al., 1968; Erhardt & Meyer-Bahlburg, 1981; Hampson, Rovet, & Altmann, 1998; Zucker et al., 1996). On balance, however, evidence suggests that differences between CAH-affected and -unaffected females is limited to the domains of childhood play behavior, aspects of gender identity, and sexuality. (For details, see the comprehensive review in Jordan-Young, 2010.)

Even within these broad domains, effects are often subtle and narrow. For example, 14 of 18 studies that examine aspects of play behavior find that CAH girls are “more masculine” on at least one dimension. However, on closer analysis, there are more negative than positive findings for most specific aspects of play. The single robust finding is that girls with CAH are more likely to play with so-called “boys’ toys” such as building blocks or vehicles, rather than so-called “girls’ toys” such as dolls or cooking implements (Berenbaum & Hines, 1992; Pasterski et al., 2005). There are no consistent differences between CAH and unaffected girls for any other aspect of play, including preference for playing with boys (Berenbaum & Snyder, 1995) or “rough-and-tumble” play (Hines & Kaufman, 1994).

Differences are also seen in gender identity, which includes one’s basic sense of self as male or female (or, rarely, as neither), and more indirect or peripheral aspects of identity such as degree of certainty, comfort, and happiness with one’s gender. While all studies to date show that the vast majority of genetic females with CAH who are reared female develop an unremarkable female gender identity, there are higher rates of male gender identity, as
well as unhappiness or dissatisfaction with female gender, in this group (Dessens et al., 2005; Gupta et al., 2006; Meyer-Bahlburg, Dolezal, Baker, Ehrhardt, & New, 2006; Meyer-Bahlburg, Gruen, & New, 1996; Sliper, Drop, Molenaar, & Keizer-Schrama, 1998).

Finally, there are differences in sexuality. From the first report on adult women with CAH, which suggested that higher numbers of (male) partners and higher libido, as well as “homosexual inclinations, the erotic response to perceptual material and the personal freedom in sex, indicate behavior more often found in males than in females” (Ehrhardt, Evers et al., 1968; 121), many reports have found “altered” sexuality in women with CAH. But contrary to narrative reviews, the contemporary pattern differs sharply from the “robust” sexual appetites and varied behaviors of the late-treated women who were the focus of early reports. Both groups show lower marriage and fertility rates, and more sexual fantasies or behaviors involving women. But women with CAH whose treatment began early in life tend to show decreased libido, lower rates of sexual function and activity, and reduced orgasmic capacity and sexual pleasure (Dittmann, Kappes, & Kappes, 1992; Dittmann et al., 1990; Hines, Brook, & Conway, 2004; Kuhnle & Bullinger, 1997; Meyer-Bahlburg, 2001; Meyer-Bahlburg & Dolezal, 2008; Mulaikal, Migeon, & Rock, 1957; Wisniewski, Malouf, & Gearth, 2004; Zucker et al., 1996).

Toward a “Whole Body”, context-sensitive account

Below, I review four aspects of developmental context that are systematically omitted in the brain organization literature on CAH (though the evidence is readily available from medical reviews and other scientific literature that describes CAH in contexts where the focus is not on effects of early androgens on the brain): 1) non-genital physiological variables that are affected in CAH; 2) intensive medical interventions and surveillance; 3) the direct effects (i.e., not indirect via gender socialization) of atypical genitalia; and 4) labeling effects of the diagnosis of CAH that prime expectations of masculinization.

1. CAH affects multiple aspects of physical morphology that are easily observable by others, and are a basis on which the growing child can compare herself. Girls with CAH are born longer, but by late childhood, are substantially shorter and heavier than average (Speiser & White, 2003; Stikkelpbroek, Hermus, Braat, & Otten, 2003; Stikkelpbroek, Oven, Van der Wilt, Hermus, & Otten, 2003): among patients at 18 centers (in USA, France, Italy, Czechoslovakia, Australia, UK, Spain, Germany, Finland, and Canada), average adult height was 1.4 standard deviations below the population mean (Eugster et al., 2001). Mulaikal et al. (1987) found that women with salt-wasting CAH had a mean height at the 25th percentile for normal women, and those with the simple virilizing form were at just the 10th percentile (p. 179). A recent large study (n = 89) of children and adolescents with CAH found a more than 7-fold increase in the rate of obesity (16.8% vs. 2.27% expected, based on general population data) (Volk, Simm, Beier, & Dori, 2006); even well-controlled CAH is associated with significant risk of obesity (Cornean, Hindmarsh, & Brook, 1998). A recent large study found that, compared to matched controls, adults with CAH had high rates of metabolic abnormalities, “including obesity (41%), hypercholesterolemia (46%), insulin resistance (25%), osteopenia (40%), and osteoporosis (7%)”; patients reported significantly more impairments across all eight domains of a standard measure of subjective health status (Arlt, Willis, Wild, Krone, & et al, 2010; 5110).

Hormone control is difficult in CAH, and common characteristics related to androgen elevation in both classical and non-classical CAH include severe, cystic acne, hirsutism, male-pattern (frontal) baldness, and failure to menstruate or irregular menstrual periods (Lin-Su, Nimkarn, & New, 2008; White & Speiser, 2000). These physical correlates of CAH may persist even if medical management has been excellent since infancy, yet brain organization studies universally ignore such physical conditions. This is true even for studies reported by investigators who elsewhere address the broad range of physical problems associated with CAH. For example, Maria New, a clinician who runs a large CAH clinic and has co-authored multiple psychobehavioral studies, notes that women with non-classical CAH commonly report “a history of discomfort and social stress related to their pretreatment experiences with androgen-dependent signs such as acne, hirsutism, and conception difficulties” (New, 2006; 4208). Yet in brain organization reports that New has co-authored, these factors are not entertained as possibly contributing to psychosexuality (e.g., Meyer-Bahlburg & Dolezal, 2008; Meyer-Bahlburg et al., 2006).

From the perspective of gendered embodiment, these outward signs of CAH are profoundly important. As Willis, Miller, and Wyn (2001) note, “Masculinity and femininity ascribe social meanings to the biological differences and similarities between the sexes” (1165), and this has salience for how intra-sex variations in physique are interpreted, as well. Once a Western, white, and especially upper-class norm, thinness has arguably become a nearly-global ideal for female beauty (Bordo, 1993), and “multiple cultures of femininity” discriminate this ideal through a “plurality of symbolic representations” that link ideal feminine beauty to thinness (Eckermann, 2009; 10). Further, as Toerien et al. (2005) explain in contextualizing their study of “mundane” production of femininity through depilation, “there is strong evidence of a widespread symbolic association between body hair — or its absence — and ideal gender: to have a hairy body is a sign of masculinity; to have a hairless one, a sign of femininity” (399).

Physical effects of CAH may cause affected girls and women to be perceived as “masculine” in two interrelated ways: first, by displaying physical traits that are more prevalent in males (e.g., male-pattern baldness, but heavier body and facial hair); and second, by displaying traits that are at odds with conventional norms of female attractiveness — regardless of whether these traits are more or less common in males (e.g., higher body mass relative to height, extreme shortness). Moreover, even traits that are read as “unattractive” in males (e.g., acne) do not detract from masculinity, though failure to conform to conventional ideals of female beauty is perceived as unfeminine, because attractiveness is central to normative femininity.

To contextualize the effect of subtle differences in appearance between girls with CAH and the unaffected relatives to whom they are usually compared, consider two studies in which observers rated girls with gender identity disorder as less attractive and more “masculine” in appearance than other girls (Fridell, Zucker, Bradley, & Maing, 1996; McDermid, Zucker, Bradley, & Maing, 1998). Fridell et al. emphasized that masculine-identified girls may prompt these ratings by adopting “masculine” clothing and hair styles, but also acknowledged that “From a transactional perspective ... one could argue that the masculine appearance of at least some of these girls would have a marked effect on their social interactions and self-representations and thus contribute to the consolidation of a masculine gender identity” (Fridell et al., 1996; 27). It is particularly notable that the senior investigator on these studies, Kenneth Zucker, has authored a number of brain organization reports on CAH, but has never presented a similarly transactional explanation for the slightly higher “masculine” gender identity observed in girls and women with CAH. Importantly, this is not to propose that there is something objectively “unattractive” about girls and women with CAH. The point is, instead, precisely the sort of transaction that Fridell et al. propose. Because dominant, white Western culture (which is likely to be the primary reference point for most girls with CAH, especially in these U.S. and European studies) prizes features
such as thinness, hairlessness, long legs, and light bone structure as especially “feminine,” girls without these features may be perceived by others and themselves as somewhat masculine, especially when expectations of masculinity have been primed by a diagnosis of CAH.

Mood problems, especially depression, anxiety and affective distress, are also often reported in boys and girls with CAH (Johannsen, Ripa, Mortensen, & Main, 2006; Kuhnle, Bullinger, & Schwarz, 1995). In addition to the stigma of the condition and potential trauma related to treatment (discussed below), mood problems may be traced to imbalances with steroid hormones, including cortisol, and the cortisol precursors adrenocorticotropic hormone (ACTH) and corticotropin releasing hormone (CRH), a neurotransmitter centrally involved in endocrine, autonomic, immune, and behavioral responses to stress. Charmandari et al. (2004) found hormone and mood atypicalities even in non-clinically affected carriers of the genetic mutation (21-hydroxylase deficiency) that causes most cases of CAH. They also noted that cortisol is frequently undersuppressed in CAH, and “chronic mild hypocortisolism has been associated with several human disorders, such as fibromyalgia and chronic fatigue syndrome, as well as atypical depression and the postpartum period” (223). Jacobs, Edelheit, Coleman, and Verzerog (1996) reported twelve patients with non-classical CAH who had refractory anxiety disorders secondary to elevated dehydroepiandrosterone (DHEAS) (a hormone also involved in cortisol synthesis). Perhaps because these neuroendocrine effects are not obviously related to gender, brain organization studies systematically ignore them. Yet anxiety, depression, and other mood disruptions could plausibly explain some CAH-related issues that are interpreted as gender-related in the context of comparisons between CAH-affected and unaffected women, including dissatisfaction with or anxiety related to gender identity, and low libido and/or orgasmic capacity.

Just one brain organization study has examined the effect of chronic illness itself on psychosexuality in CAH. Sluijer (1984) found that both girls with CAH and girls with diabetes scored as more “masculine” than healthy controls on a gender scale (though this was more pronounced among girls with CAH), and concluded that “being sick plays a role” in gendered behavior.

2. Women and girls with CAH are subject to intensive, life-long medical and psychiatric intervention, including both invasive physical procedures and persistent scrutiny and heightened concern about gender and sexuality. Some 85% or more of girls with classical CAH undergo surgery meant to “feminize” the genitalia. The norm is a minimum of two surgeries, most commonly both in infancy and in adolescence and/or adulthood, either to “finish normalization” or to correct problems from earlier surgery(ies), including vaginal stenosis, fistulas, and incontinence (Crouch, Laio, Woodhouse, Conway, & Creighton, 2008; Crouch & Creighton, 2007; Mulaikal et al., 1987; May, Boyle, & Grant, 1996; Stikkelenbroeck, Hermus et al., 2003; Stikkelenbroeck, Oyen et al., 2003). The vast majority of women and girls with CAH in brain organization studies have had clitoral surgeries (e.g., Meyer-Bahlburg et al., 2006; Pasterski et al., 2005; Servin et al., 2003); these include total amputation of the clitoris (clitoridectomies), clitoral “reduction,” a surgery that involves removal of the erectile tissue, and clitoral “resection”, in which clitoral structures are “tucked” under the pubis. While clitoral recession is now less frequent, because it is associated with painful clitoral erections, a shocking number of women and girls with CAH have had total amputation, even in fairly recent studies (e.g., May et al., 1996; Riepe et al., 2002). Data suggest that all genital surgeries leave most women with nerve damage, impairing sensitivity and orgasmic capability, sometimes severely. Crouch et al. (2008) examined sexual function and genital sensitivity in women with CAH who had surgery (n = 28), women with CAH without surgery (n = 4) and normal controls (n = 10). They found no sensitivity differences in the upper vagina, where surgery had not been performed in any women, but found marked impairment in sensitivity in all areas where “feminizing” surgery had been done; they also found a linear relationship between impairment of sensitivity and the severity of sexual difficulties.

Medical visits every three or four months are considered necessary to monitor hormone levels and response to treatment in children and adolescents with CAH (Riepe et al., 2002; Rivkees, 2002). Medical monitoring involves regular inspection of the genitals, because clitoral growth or lack thereof is one sign of “good” hormonal management; genital inspection may also determine whether such treatments as vaginal dilation have been successful in lengthening the vagina or keeping it open after surgery. The routine genital inspection at clinical visits “has been likened to sexual abuse” (Berenbaum, 2004; 4) and “women with CAH often remember childhood visits to their physician as highly intrusive” (Speiser et al., 2010; 4149). In one follow-up study, women with CAH “used a language of ‘rape’, ‘invasion’ and ‘violation’ when talking about vaginal examinations and other procedures carried out during visits to pediatric and adult clinics” (May et al., 1996). In interviews with medical anthropologist Katrina Karkazis (2008; 190–191), girls with CAH, as well as their parents, frequently described medical scrutiny of genitals as “intrusive and dehumanizing”.

While overall psychological adjustment seems to be acceptable (Berenbaum, Bryk, Duck, & Resnick, 2004; Kleinemeier et al., 2010; Yates et al., 1999), girls and women with CAH have high rates of negative body image (Kuhnle & Bullinger, 1997) and anxiety (Mueller et al., 2010), both of which are plausibly related to sexualized medical surveillance (the latter may also be a result of cortisol disruption). May et al. (1996) offer particularly poignant data on how medical interventions may affect women’s relationships to their bodies. Fewer than half of women with CAH reported ever masturbating, compared with three-quarters of women with diabetes; women with CAH “commonly spoke about masturbation as a necessary medical procedure rather than primarily as a sexual activity” (484).

Perhaps because they do not engage the hypothesis of brain androgenization, these studies of sexual activity and function among women with CAH that consider the physical and psychological sequelae of the management process itself are never mentioned in the brain organization literature. In fact, Iain Morland (2011), points out the irony that genital surgeries in infants are rationalized as “at once life-changing (in imprinting gender) and not life-changing at all (in having no impact other than the imprinting of gender)” (159). In those few studies where researchers do consider the effect of treatment on psychosexual outcomes in CAH, they routinely assume that treatment can only reduce differences between patients and healthy comparison groups. In the words of Gastaud et al. (2007), “Despite expert medical and surgical care by physicians dedicated to this rare disease, women with CAH still suffer major limitations in their sexual function and reproductive life” (1391). Yet, given that 100% of the women with classical CAH in this study had genital surgeries, it would seem more reasonable to frame the poor outcomes as because of the medical care, rather than despite it. A particularly compelling argument for this interpretation comes from comparing sexual function among women who have been treated from infancy with those whose treatment began in adolescence or adulthood (e.g., Ehhardt, Epstein et al., 1968). Late-treated women reported more male partners, higher libido, a lower threshold to arousal and more sexual versatility than the investigators anticipated (though the “expected” norms to which women
were compared were arguably dated and conservative). Given that both late-treated and early-treated women have similar prenatal histories, it seems prudent to suppose the discrepancy is explained by factors that are different between these groups: early diagnosis, early surgeries, and lifelong suppression of androgens among the early-treated women.

3. Atypical genitalia have direct effects on sexuality (not only indirect effects via gender socialization). Remarkably, genital morphology is routinely overlooked when investigators report on CAH-affected women's sexuality in the context of brain organization research. Early reports, for example, suggested that women with CAH were sexually “versatile,” because they reported trying many sexual positions. Investigators took this to reflect brain masculinization (e.g., Ehrhardt, Evers et al., 1968; Masica, Money, & Ehrhardt, 1971), apparently never considering that a short or inflexible vagina might lead a woman to experiment in order to find more comforting or pleasurable positions.

The most profound difference in sexuality between women with CAH and unaffected women is that the former have very low rates of sexual activity and partnerships across the board, as well as lower levels of sexual arousability, genital sensation, and orgasmic capacity. The majority of adult women with classical CAH have impaired clitoral sensation, and one-third to two-thirds have a vaginal opening that is not adequately large or flexible to permit heterosexual intercourse (Crouch et al., 2008; Gaudat et al., 2007). One of the few long-term outcome studies to report on multiple components of sexual function indicates that, for most women with CAH, heterosexual intercourse is painful: “37% (13 of 35) said they never had heterosexual intercourse with vaginal penetration” and 81% of the women who did have heterosexual intercourse experienced pain during penetration (Gaudat et al., 2007; 1393). Women born with the most severely affected genitalia reported severe impairment in all aspects of sexual function (e.g., lubrication, orgasm, satisfaction with sex acts), but even the women who had less severely affected genitalia reported significantly higher rates of vaginal pain, and less lubrication and orgasm, than the healthy women to whom they were compared (Gaudat et al., 2007; 1394). Another study found similar results, even when CAH women were compared not to healthy controls, but to women with early-diagnosed diabetes mellitus, which may also impair sexual function: women with CAH had less sexual experience, and reported higher levels of penetration difficulties, pain, and lubrication problems than the diabetic women (May et al., 1996).

Women with CAH are widely described as more likely to be bisexual or lesbian than other women. Interestingly, though, most studies show rates of same-sex orientation among women with CAH that are higher than comparison groups (typically unaffected female relatives) (Gaudat et al., 2007; Hines et al., 2004; May et al., 1996; Meyer-Bahlburg & Dolezal, 2008; Zucker et al., 1996), but not higher than women in the general population (see Jorn, Dear, Rodgers, & Christensen, 2003; Savin-Williams, 2006; Sell, Wells, & Wypij, 1995). The possible exception is women who were initially assigned as male (Meyer-Bahlburg et al., 2008). Where women with CAH do report increased same-sex orientation compared to general population rates, this is mostly due to fantasy or attraction, while rates of actual same-sex behavior are only slightly elevated, if at all. In a very large study that is now more than 20 years old, Mulaikal et al. (1987) found that sexual orientation and activity more closely related to women's vaginal condition than to the degree of prenatal androgen (p. 180). Given that heterosexuals in the contemporary U.S. context interpret “having sex” to be synonymous with penile-vaginal intercourse (Bogart, Cecil, Wagstaff, Pinkerton, & Abramson, 2000; Sanders & Reinsch, 1999), it does not seem surprising that women who, as a group, have extremely low rates of “typical” vaginal size and function, would also have somewhat increased rates of at least fantasizing about female partners, where sexual intimacy would not necessarily hinge on the depth, flexibility, and lubrication of the vagina. Yet by routinely interpreting same-sex attractions as well as lower rates of penile-vaginal intercourse as evidence of “brain masculinization”, brain organization researchers take for granted that the normal and natural state of affairs is both that women will be exclusively attracted to men, and that heterosexual eroticism will center on penile-vaginal intercourse.

4. Diagnosis with CAH activates expectations of masculinization that likely affect both development and assessments of gender and sexuality in CAH-affected women and girls. Feminist biologists and other critics have noted for decades that being born with unusual genitalia initiates an enormously unusual sequence of developmental events for girls with CAH (Bleier, 1984; Longino & Doell, 1983). Scientists conducting brain organization studies continue to give short shrift to how clinicians' and parents' concerns about masculinization may affect development (e.g., Hines, 2010). Most articles devote no more than two to three lines of text to the issue, and none have systematically investigated how labeling or priming effects associated with CAH diagnosis may affect parental perception of “masculine” characteristics, nor girls' actual developing behavior. These omissions are important, and I have explored them elsewhere (Jordan-Young, 2010). Here, it must suffice to note that the connection among perceived status/perceived attributes/actual behavior, and assessments of behavior constitute a “looping” process. Research on how gender as a status triggers this sort of perception-behavior-evaluation loop generally focuses on male-female differences, but we can easily apply the observation to girls with CAH, who are seen as having a “diminished” femininity, or rather, are suspected of being masculine. Since brain organization theory was introduced, masculine traits and behavior among girls and women with CAH have been predicted by scientists and clinicians, and these expectations have been communicated to parents, as well as to women and girls with CAH themselves. Importantly, the evaluation of traits and behavior in research on CAH is almost entirely conducted by people who are aware of both the theory and the subjects' status as affected or unaffected by CAH, with “blind” ratings and observations the very rare exception. Thus, the diagnosis of CAH may be thought of as a lifelong “frame” through which clinicians, parents, and girls and women with CAH make sense of gender and sexuality, which likely affects both the development of gender and sexuality, and the way that individuals' traits and behaviors are perceived and reported.

Evidence for the idea that brain organization theory operates as an interpretive frame is the uncanny tendency for investigators to interpret any differences between women with CAH and unaffected women as “masculinization,” even when that interpretation requires reversal in the criteria for measuring masculine versus feminine sexuality. As noted above, investigators initially found that women with CAH had higher levels of libido, masturbation, and number of partners, as well as other aspects of sexuality that were at that time considered “masculine”, but more recent studies have consistently found lower rates of virtually all aspects of sexuality that were initially considered masculine. This cohort difference has gone unremarked in the literature, perhaps because of a dramatic shift in definitions for what constitutes “masculinization.” In recent reports, lower levels of libido, less frequent and varied sexual activity, fewer and less lasting sexual partnerships, and reduced fertility among women with CAH are often interpreted, remarkably, as indicating brain masculinization (Meyer-Bahlburg, 1999; Zucker et al., 1996; Zucker et al., 2004). This characterization is hard to justify. For example, masturbation, while certainly not the exclusive domain of males (as it was framed in the early studies of CAH) is reliably reported to be practiced more frequently by males than females in the broader populations from which these CAH samples
are drawn (Gerressu, Mercer, Graham, Wellings, & Johnson, 2008; Laumann, Gagnon, Michael, & Michaels, 1994).

Conclusion

Studies of CAH are routinely invoked as forming the strongest pillar of evidence supporting the theory that steroid hormones in utero shape “brain gender” in humans. In turn, sex differences in career interests, domestic roles, learning styles, sexuality, and sex-typical patterns of mental health and disease are justified to some degree (that varies by the writer) as “natural” (Berenbaum & Resnick, 2007; Hines, 2010; Sax, 2005). Yet this literature is characterized by tunnel vision — researchers focus on prenatal androgen exposures, but ignore morphologic and metabolic differences between CAH-affected and unaffected groups, intensive medical intervention and monitoring, the direct and indirect effects of atypical genitals, and the way that a CAH diagnosis may prime clinicians, parents, and patients themselves to expect and perceive masculinization.

As an interpretive “frame”, brain organization theory encourages researchers to push aside more observable and proximate factors — biological, social, and the iterative interactions among these — that impinge upon the development of gender and sexual function, in favor of the presumption of early hormonal influence. This frame covers gaps and inconsistencies in the data that presumably indicate brain organization by prenatal hormones in women with CAH, obscuring reversals in key definitions and inconsistencies between treatment cohorts. Such empirical inconsistencies destabilize the scientific status of brain organization theory. The consequences are not just that brain organization theory continues to be given more weight than it merits. These studies, with their fixation on early hormonal effects, contribute to a systematic disregard for how medical intervention affects girls with CAH, and others with intersex conditions who are subjected to cosmetic, but medically-unnecessary, genital surgeries. Further, they contribute to an environment in which parents, as well as girls and women with CAH, are subject to heightened concerns about inadequate femininity, and pressured toward outmoded and questionable views of “normal” gender and sexuality.

While it may be accurate to describe certain physical functions such as nerve conductivity or thermal sensitivity as “impaired”, the same cannot be said of gender or sexuality. In Morland’s (2011) words, “From a feminist standpoint that regards gender as a social doing, one cannot assume gender to be present in a newborn and susceptible to surgical alteration, diminution, or loss” (160). Likewise, from the perspective that sexuality (like gender) is emergent and transactional rather than an “innate” property of bodies or individual psyches, I have ventured to explore how physical characteristics like genital morphology might affect sexual orientation or activities, without pathologizing either atypical physicality or atypical psychosexuality. Thus, this paper has not argued that the “natural” or “innate” gender and sexuality of people with CAH is damaged by surgeries and other interventions, nor by labeling and priming effects, nor the interaction with complex physiologic characteristics that are atypical in this condition. The point, rather, is that all of these factors matter. Only by willful ignorance, fueled by a stubborn commitment to brain organization theory and perhaps also to the worldview that it seems to justify (see Proctor, 2008), can they continue to be disregarded.

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References


