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An Alternative Paradigm for the Psychosocial Management of Patients with DSD

David E. Sandberg PhD

Division of Child Behavioral Health

Department of Pediatrics & Communicable Diseases



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Management of DSD

Changing Practices



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“True Sex” versus “Optimal Gender”



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True Sex: until mid-1950s medical management guided by belief that individual's true sex revealed through examination of internal anatomy.

Optimal Gender: gender assignment considers multiple aspects of outcome, most prominently potential for complete sexual functioning



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Optimal Gender Policy



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Designed to address shortcomings of gender assignment based on one criterion (eg, gonads, chromosomes)

Small window of gender flexibility (~18 months of age)

Perform “normalizing” genital surgery early in life:

- To remove any parental doubt about the sex of the child
- To promote stable gender identity

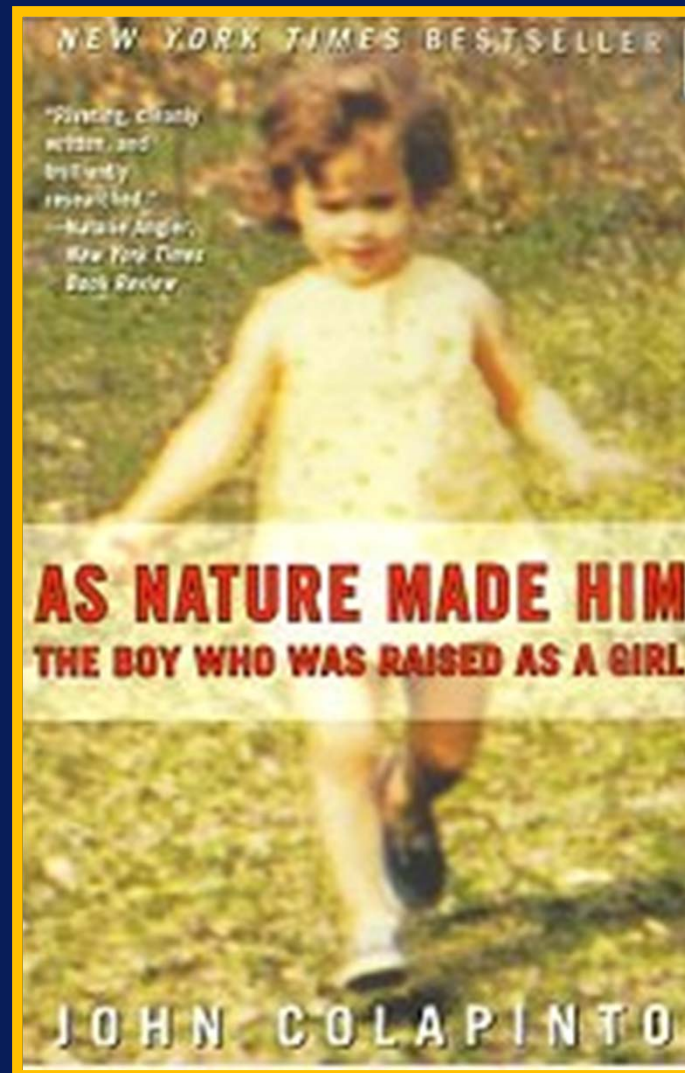


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Nature Trumps Nurture?



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2000



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Nature Trumps Nurture?



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.... but, perhaps not

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**Experiment of Nurture: Ablatio Penis at 2 Months, Sex Reassignment at
7 Months, and a Psychosexual Follow-up in Young Adulthood**

Susan J. Bradley, Gillian D. Oliver, Avinoam B. Chernick and Kenneth J. Zucker

Pediatrics 1998;102:e9

DOI: 10.1542/peds.102.1.e9



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Chicago Consensus Conference 2005



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Consensus Statement on Management of Intersex Disorders

Peter A. Lee, Christopher P. Houk, S. Faisal Ahmed, Ieuan A. Hughes and in collaboration with the participants in the International Consensus Conference on Intersex organized by the Lawson Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology

Pediatrics 2006;118:e488-e500

DOI: 10.1542/peds.2006-0738



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Example of a DSD Classification



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Sex Chromosome DSD

46,XY DSD

46,XX DSD

- | | | |
|-----------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------|
| A. 45,X (Turner syndrome and variants) | A. Disorders of gonadal (testicular) development (e.g., complete gonadal dysgenesis (Swyer syndrome)) | A. Disorders of gonadal (ovarian) development (e.g., ovotestic DSD; testis DSD (SRY+)) |
| B. 47,XXY (Klinefelter syndrome and variants) | B. Disorders in hormone synthesis or action (e.g., andro biosynth defect (17-HSD defic, 5 α -reductase defic); defect in andro action (cAIS, pAIS)) | B. Androgen excess (e.g., fetal (21-OH deficiency, 11-OH) deficiency) |
| C. 45,X/46,XY (mixed gonadal dysgenesis) | C. Other (e.g., severe hypospadias, cloacal exstrophy) | C. Other (e.g., cloacal exstrophy, vaginal atresia) |
| D. 46,XX/46,XY (e.g., ovo-testicular DSD) | | |



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DSD: Moving in a New Direction



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- Conference a consequence of advances in diagnosis, surgical techniques, and recognition that not all involved satisfied with the model of care
- Identifies outcomes beyond psychosexual development (gender identity, gender role, sexual orientation) as important
 - e.g., quality of social relationships, health-related quality of life
- Acknowledgement that social factors can modify outcomes



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What do we know about CAH?



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- Most common 46,XX DSD and most extensively studied from psychological outcomes standpoint
- Autosomal recessive mode of inheritance
- Psychological outcome studies predominantly of affected girls and women
 - opportunity to test hormonal theories of psychosexual differentiation in humans
 - however, not typically examined in context of broader developmental literature or research on pediatric chronic illness



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Systematic Review: Data



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Domain	Subdomain	% Studies
<i>Psychosexual Differentiation</i>	gender identity, gender role, sexual orientation	68%
<i>Psychological Adaptation</i>	self-concept, behav/emot funct, psychopath, HRQoL	31%
<i>Sexuality</i>	sexual function/activity	25%
<i>Social Adaptation</i>	social funct. cohab/marriage	25%
<i>Cognitive Function</i>		23%
<i>Reproduction</i>	fertility status, conception	10%
<i>Other</i>		8%
<i>Education/Occupation</i>		7%



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What do we know about CAH?

psychological endpoints



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Psychosexual Differentiation

– Gender Identity

- predominantly identified as girls and women with minority self-reassigning later in life

– Gender Role

- girls/women exhibit more masculine/less feminine behaviors/interests

– Sexual Orientation

- higher prevalence of non-heterosexual orientation



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CAH: Psychological Endpoints



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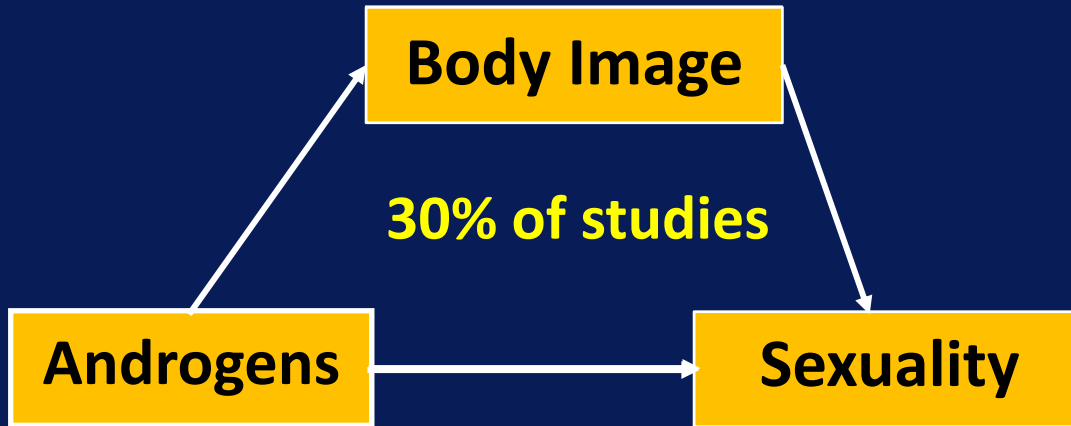
Dominant Conceptual Model



73 of 98 studies (74%)



Mediation and Moderation

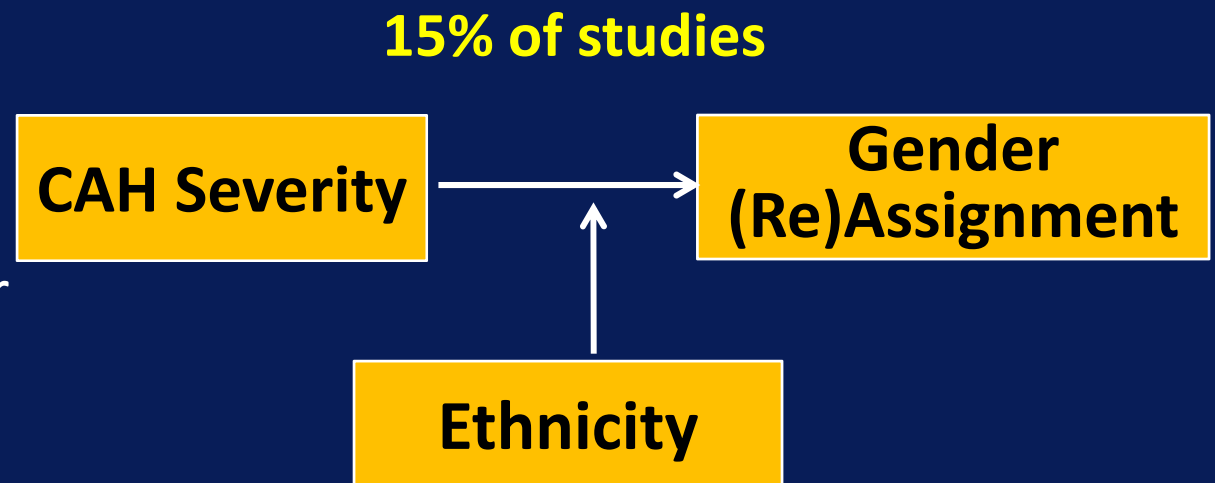


Mediation

- Predictor has both a direct and indirect relationship to an outcome
- Factor X represents mechanism by which effect occurs

Moderation

- Direct relationship between predictor and outcome
- Factor X determines under what circumstances effect occurs





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Summary & Conclusions



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- Categorization of conceptual/theoretical models in CAH (**and other DSD**) suggests prenatal androgen exposure as primary predictor of psychological outcomes
- Relatively few studies designed to examine mediating or moderating influences of postnatal experiences on psychological outcomes



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Future Directions



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Study of postnatal (e.g., surgical/social/intra-psychic) variables mediating and moderating the relationships between medical diagnosis and quality of life outcomes will inform:

- Development of theoretical models accounting for variability in psychosexual and other psychological outcomes
- Clinical strategies to modify the influences of DSD on quality of life outcomes.



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