

Children's Mercy Kansas City

## SHARE @ Children's Mercy

---

Research Days

GME Research Days 2020

---

May 15th, 12:45 PM - 1:00 PM

### Change in Sex of Rearing in Individuals with Disorders of Sex Development

Michelle Knoll

Children's Mercy Hospital, mmknoll@cmh.edu

Follow this and additional works at: <https://scholarlyexchange.childrensmercy.org/researchdays>



Part of the [Endocrine System Diseases Commons](#), [Female Urogenital Diseases and Pregnancy Complications Commons](#), [Male Urogenital Diseases Commons](#), and the [Pediatrics Commons](#)

---

Knoll, Michelle, "Change in Sex of Rearing in Individuals with Disorders of Sex Development" (2020). *Research Days*. 4.

[https://scholarlyexchange.childrensmercy.org/researchdays/GME\\_Research\\_Days\\_2020/researchday5/4](https://scholarlyexchange.childrensmercy.org/researchdays/GME_Research_Days_2020/researchday5/4)

This Oral Presentation is brought to you for free and open access by the CONFERENCES, EVENTS, GRAND ROUNDS at SHARE @ Children's Mercy. It has been accepted for inclusion in Research Days by an authorized administrator of SHARE @ Children's Mercy. For more information, please contact [library@cmh.edu](mailto:library@cmh.edu).

# Change in Sex of Rearing in Individuals with Disorders of Sex Development

**Submitting/Presenting Author (must be a trainee):** Michelle Knoll, MD

**Primary Email Address:** mmknoll@cmh.edu

**Resident/Psychology Intern ( $\leq$  1 month of dedicated research time)**

**Resident/Ph.D/post graduate ( $>$  1 month of dedicated research time)**

**Fellow**

**Primary Mentor (one name only):** Jill Jacobson, MD

**Other authors/contributors involved in project:** Anna Egan, PhD; Julie Strickland, MD; John Gatti, MD; Joel Koenig, MD

**IRB Number:** 15080356

**Describe role of Submitting/Presenting Trainee in this project (limit 150 words):**

Dr. Knoll conceptualized the project with the assistance of Dr. Jacobson and Dr. Egan, collected all the data, did preliminary data analysis, and wrote the abstract.

**Background, Objectives/Goal, Methods/Design, Results, Conclusions limited to 500 words**

## **Background:**

Differences of Sex Development (DSDs) encompass variations in formation of internal or external sex characteristics. Historically, sex assignment in children with DSDs depended on phenotype, and gender was thought to be malleable. Attention has recently shifted toward reducing gender dysphoria and preserving fertility potential in individuals with DSDs.

## **Objectives/Goal:**

Our objective in this study was to determine the rates of sex reassignment and gender dysphoria in our patients with DSDs.

## **Methods/Design:**

A chart review was done on all patients seen in GUIDE clinic between April 2008 and June 2019 to determine rates of sex reassignment and gender dysphoria. Two hundred patients were seen: 23 were found to not have DSDs; 61 were 46,XX; 66 were 46,XY; 31 had a sex chromosome DSD; 5 had gonadal dysgenesis without known chromosome mosaicism; and 14 had syndromic genital atypia. Mean age of follow-up is 8.77 years.

## **Results:**

Only 2 patients underwent sex reassignment at our institution. One was assigned male at birth, but was found to be 46,XX with 21-hydroxylase deficiency and was reassigned female at 1 month of age. The second was assigned male at birth and was found to be 46,XY with an NR5A1 variant. Sex was reassigned female at two months of age. Two additional patients had a sex reassignment outside our institution. One was born abroad and assigned male at birth. The patient was found to be 46,XY with an NR5A1 variant, and was reassigned female. The second patient was assigned male at birth, but was found to be 46,XX with P450 oxidoreductase deficiency. The parents changed the sex of rearing to female at 19 months of age. To date, none have signs of gender dysphoria.

A total of three patients experienced gender dysphoria and underwent transition. The first had genital ambiguity with sex chromosome mosaicism in the gonads and was assigned female at birth. We held care conferences with the family and discussed the possibility of gender dysphoria. At age 3, the patient declared that he was male, and parents socially transitioned him at that time. Two were assigned female after receiving the diagnosis of 46,XX 21-hydroxylase deficiency. Both declared male gender identity later, one at 12.5 years of age, and one at 20.5 years of age.

**Conclusions:**

Whereas our patient population is still relatively young, it is reassuring that the overall rate of gender dysphoria is low. The rate in patients with CAH is similar to previous reports in the literature. Careful attention to sex assignment in early childhood may reduce the rates of gender dysphoria in children with DSDs.