

University of Michigan Initiative on Rare Disease Research

Disorders of Sex Development (DSD) Research and Quality Improvement Symposium



David E Sandberg, PhD
Director, Child Behavioral Health, Department of Pediatrics & Communicable Diseases
University of Michigan, Ann Arbor, Michigan

Anthony J Ascianto, MA, MPH
Executive Director, Accord Alliance
www.accordalliance.org



INTRODUCTION

Disorders of sex development (DSD)[†] are congenital conditions in which chromosomal, gonadal, or anatomic sex development is atypical. For families, the birth of a child with a DSD, and attendant uncertainty about the child's gender and future psychological and sexual development, is believed to be extraordinarily stressful. For healthcare professionals, genital ambiguity and discordance genotype, gonads, and anatomy can be the most challenging aspect of an already complex medical condition for which long-standing controversy over the most appropriate model of care exists.

SYMPOSIUM GOAL

To create a vision for research on caring for persons affected by DSD to guide recommendations for an interdisciplinary research and collaborative learning agenda.

SPECIFIC AIMS

- Develop consensus among invited stakeholders on focus areas for research and collaborative learning to drive improvements in DSD healthcare and outcomes.
- Develop recommendations that will generate interest and support for initiatives to close the gap on the evidence and information available to providers and persons affected by DSD.
- Establish a network of researchers, providers, and patients and families with a stake in improving treatment and care for DSD that will work together to champion continued research and learning.

SYMPOSIUM INVITEES

By invitation and referral, co-investigators at 12 sites across the US involved in NIH/NICHD R01 HD053637 (Quality of Life in Intersexuality: A Pediatric Clinical and Research Tool; PI: Sandberg), patient and family members, patient advocacy groups, governmental agency representatives, and content experts representing understudied areas in DSD, including:

- **Providers:** pediatric endocrinology, urology, genetics, social work, psychology, child life, nursing;
- **Researchers:** molecular genetics, urology, endocrinology, quality of life outcomes research, cost-effectiveness research;
- **Content experts:** shared decision-making, psychological outcomes in DSD research, informed consent for research/care, and team-based care;
- **Affected persons:** unaffiliated adults with DSD;
- **Parents of affected persons:** unaffiliated with advocacy organizations and representatives of advocacy organizations
- **DSD advocacy groups:**
 - Hypospadias & Epispadias Association <http://heainfo.org>
 - The MAGIC Foundation <http://www.magicfoundation.org>
 - Androgen Insensitivity Syndrome Support Group <http://www.indiana.edu/~ais/html/home.html>
 - Turner Syndrome Society of the USA <http://www.turner-syndrome-usa.org>
- **Bioethics and Legal Experts:**
 - Advocates for Informed Choice <http://www.iiclaw.org>
 - Former Associate Vice President and Deputy General Counsel, University of Michigan
- **Michigan Department of Community Health:** State of Michigan Department of Community Health Newborn Screening Program
- **National Institutes of Health:** Director of the Office of Rare Diseases Research <http://rarediseases.info.nih.gov>



Symposium participants gather for a group photo
Palmer Commons, University of Michigan, Ann Arbor

SYMPOSIUM DESIGN AND FACILITATION

The Symposium was held at the University of Michigan on April 1-3, 2009. Program organization and session facilitation was a collaborative effort between the **University of Michigan** and **Accord Alliance**, a not-for-profit dedicated to promoting comprehensive and integrated approaches to care that enhance the health and well-being of people and families affected by DSD by fostering collaboration among all stakeholders.

Two group facilitation techniques were used in combination to accommodate participant backgrounds and varying perspectives and encourage active discussion and consensus building:

❖ Conversation Café discussions

A semi-structured process for engaging large numbers of people with diverse backgrounds and perspectives in meaningful discussion. Conversation Café are used to increase personal and group knowledge and awareness around important issues.

1. Content experts introduced topics relative to DSD research and outcomes.
2. Participants joined pre-assigned small groups (6-8 people) to explore and expand on each topic in rapid (20-minute) discussions.
3. Small groups presented discussion summaries to the larger group. Facilitators roamed among groups to help discussions remain focused.

➤ Learn more about Conversation Café: www.conversationcafe.org

❖ Open Space Technology workshops

A dynamic semi-structured process for meeting facilitation that functions "...best when the work to be done is complex, the people and ideas involved are diverse, and the passion for resolution (and potential for conflict) are high..."

1. As a warm-up exercise, a rapid-panel discussion was conducted; 6 participants from distinct stakeholder groups summarized themes and offered commentary on Conversation Café topics.
2. Small-group workshops were organized around research and outcomes-focused topics nominated and rank-ordered by participants.
3. Participants served as workshop leads for those topics ranked "most important." Participants chose which sessions to attend, and were able to move between sessions at will. Sessions were not time-limited.
4. Meeting facilitators roamed among groups to guide discussions towards recommendations for future inquiry; scribes recorded discussions and themes across the small groups.

➤ Learn more about Open Space: www.openspaceworld.org

The Symposium concluded with a working lunch and facilitated wrap-up discussion coalescing on prominent themes and potential funding mechanisms to support recommendations aligned with research and quality improvement.

DISCUSSION TOPICS

▪ Opening Remarks

Stephen C. Groft, PharmD, Director, Office of Rare Diseases Research, National Institute of Health

▪ Conversation Café topics

• Psychosocial outcomes in DSD research

Tom Mazur, PsyD, Women and Children's Hospital of Buffalo and the University at Buffalo

• Informed consent for research and care

Anne Tamar-Mattis, JD, Executive Director, Advocates for Informed Choice (AIC)

• Shared decision-making

Laura A. Siminoff, PhD, Professor and Chair, Department of Social and Behavioral Health, School of Medicine, Virginia Commonwealth University

• Team-base care

Helen Sharp, PhD, CCC-SLP, Assistant Professor, Western Michigan University

▪ Sample Open Space workshop topics

• Long-term outcomes research

• for example, workshop participants included: pediatric psychology, social work, bioethics, neuroscience, and clinical decision-making

• Person and family-centered care

• for example, workshop participants included: medical information website editor, pediatric endocrinology, pediatric endocrinology nurse specialist, pediatric urology social work and DSD team lead, parent, and healthcare quality consultant

• Transferring knowledge and experience from pediatric to adult care

• Creating a research infrastructure

• Quality indicators and model of care

• Research ethics

• What research matters?

• Group communication

• Informed consent for treatment



Participants gather to review Open Space workshop summaries

REFERENCES

[†]Lee PA, Houk CP, Ahmed SF, Hughes IA, 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. Consensus statement on management of intersex disorders. *Pediatrics* 2006; 118(2):e488-e500.

ACKNOWLEDGEMENTS

Supported by a grant from the Office of the Vice President for Research, Initiative on Rare Diseases Research, University of Michigan

RESULTS

➤ Attendees represented broadest range to-date of stakeholders convening to discuss research and quality improvement in DSD

- **Providers:** pediatric endocrinology (5); pediatric urology (7); genetics (3); adolescent gynecology (1); nursing (3); social work (2); pediatric psychology (6); clinical psychology (1), child psychiatry (1), child life (1);
- **Researchers:** genetics (2), pediatric urology (5), pediatric endocrinology (1), economic evaluation of children's health interventions and valuation of health outcomes for cost-effectiveness analysis (1), health-related quality of life (5);
- **Content experts:** shared decision-making (1), psychological outcomes in DSD research (1), informed consent for research/care (1), team-based healthcare (1);
- **Affected persons** (5);
- **Parents of affected persons** (4);
- **DSD advocacy groups** (5 organizations);
- **Bioethics and Legal Experts** (3);
- **Medical writer** (1);
- **Michigan Department of Community Health** (1); and
- **National Institutes of Health** (1)

➤ Broad consensus items

- ✓ Parental understanding of and adaptation to child's diagnosis and prognosis is an important determinant of child's health-related quality of life;
- ✓ Collaborations among affected adults, patient support groups, and interdisciplinary healthcare teams will improve care and enhance outcomes;
- ✓ Outcome studies need to encompass a broader range of psychological endpoints beyond psychosexual differentiation (i.e., gender identity, gender-role, and sexual orientation);
- ✓ There is a dearth of high quality DSD educational materials available for providers working with patients and families to promote better understanding. Similarly, there are no curricula for the public to correct misunderstandings and perception of stigma associated with DSD;
- ✓ Fragmentation of care, lack of coordination, and lack of support for parents reduces quality of care. Models of integrated interdisciplinary pediatric and adult healthcare exist that could inform development of DSD teams;
- ✓ Future similar events should include participation of experts in healthcare organization and funding; aspects of healthcare delivery posited to enhance outcomes (e.g., integrated behavioral services and coordination of care are not reimbursable).

➤ Established virtual (listserve) network for participants and invited guests, providing a forum for ideas to enhance healthcare and outcomes, suggestions for focused research activity, and a resource for collaborators on potential projects (research, clinical care, and patient advocacy, etc.)

➤ Established group of collaborators who immediately began grant preparation work to fund a multi-site infrastructure necessary for hypothesis-based research on mechanisms of sex development and evidence-based care for patients/families affected by DSD.