

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES (HHS)
National Institutes of Health (NIH)
Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD)

Growing up with Disorders of Sex Development (DSD): Critical Developmental Issues for Children and Families Affected by DSD

Natcher Conference Center (Building 45), Room D
NIH main campus, Bethesda, Maryland
March 26–March 27, 2014

Meeting Summary

Approximately one of every 4,500 children is born with a variation in chromosomal, gonadal, or anatomical makeup that may make it difficult at birth to determine the infant's sex or that may not become symptomatic until puberty. These differences or DSD have important developmental and medical ramifications and may affect gender of rearing as well as health, quality of life, and family functioning during the child's infancy, childhood, adolescence, and adulthood. Yet little is known about the short- and long-term outcomes of newer surgical and hormonal interventions or decisions to forego these interventions. Even less is known about the impacts of DSD on the child during different periods of development and on the child's family; these impacts can have important implications for the timing and nature of clinical decisions as well as child, adolescent and adult health and well-being and family functioning.

The workshop gathered a multidisciplinary group of clinical and research experts who described and discussed: a) current knowledge of the etiologies of DSD as well as current clinical care and counseling for affected children and adolescents and their families; b) important knowledge gaps and research needs (including clinical infrastructure and research resources) ultimately to better inform all aspects of care for affected individuals and families; and c) specific research questions to advance the field.

The workshop experts came from several different basic and clinical research areas including developmental and clinical psychology, genetics, urology, developmental neuroscience, endocrinology, gynecology, molecular and cellular biology, bioethics, pediatrics, psychiatry, developmental psychopathology, gender studies, and social and behavioral sciences and advocacy on behalf of affected individuals and families. Despite this wide ranging multidisciplinary foundation, the workshop participants found common ground in their presentation sessions and discussion. The NICHD was also encouraged to have representatives from eleven other NIH Institutes, Centers, and Offices attend the workshop and express interest in the topic.

Representatives of affected individuals and families participated in planning the workshop and in its presentations and discussions; in addition, NICHD sought public input with a Request for

Information (RFI) and responses to the request were summarized were provided to participants before the workshop began. A summary of the RFI responses is available at <http://www.nichd.nih.gov/about/meetings/2014/Pages/032614.aspx>.

The workshop proceedings and RFI comments both emphasized needs for:

- Better basic, clinical and translational evidence on which to base DSD diagnoses, prognoses and clinical decisions
- Developmentally appropriate multidisciplinary clinical management, including patient transition to adult care
- Full, accurate, readily understandable and developmentally appropriate communication between affected individuals (including older children and adolescents), families, and clinicians to ensure that decision-making about types and timing of clinical options be as fully informed as possible
- Psychological and peer-group support for affected individuals and families
- The involvement of affected individuals and families in development and implementation of research, including research on patient-identified outcomes of importance
- Understanding and acceptance of anatomical and gender variance
- Ethical concerns

Although the NICHD has sponsored or co-sponsored previous workshops addressing DSD issues (in the early 2000s), this is the first workshop that focused exclusively on developmental issues and the transition from adolescence to adulthood. This was the also the first time that a bioethicist presented at an NICHD workshop on DSD, and that a session was included on what could learn from developmental research and clinical approaches with other unrelated, but chronic, developmental disorders such as spina bifida and congenital cranio-facial abnormalities.

At the workshop, time was set aside for each presenting expert to describe his or her suggestions for the top three research questions or needs in understanding, diagnosing, treating, and/or facilitating healthy development for individuals with DSD. These questions fell into four general themes:

- Improving the diagnosis of DSDs
 - For example but not limited to: Novel genomic approaches for increasing diagnostic accuracy
- Genitosurgery and gender assignment outcomes
 - For example but not limited to: Identification of predictive factors for more accurate gender assignment; short- and long-term outcomes of newer surgeries or when the decision is to forego surgery
- Psychosocial and functional impacts on development for individuals with DSD
 - For example but not limited to: Socio-emotional development in children with DSD and family issues; predictors of gender satisfaction or dysphoria
- Improving clinical management of DSD
 - For example but not limited to: Clinical trials for hormone supplementation or replacement; effective educational materials and methods of clinician communication

Perhaps the most striking aspect of the workshop, other than the continued limited research on DSD, was the ability of so many individuals, with so many different backgrounds, to really listen and respond thoughtfully to the variety of viewpoints and concerns on topics that have the potential to result in emotional or even contentious exchanges. This aspect of the workshop was commented upon by several participants and observers and seems to indicate that this is now the right time to address the research gaps and needs that very much require a developmental, multidisciplinary approach.

The NICHD continues to evaluate the outcomes of the workshop and the input from the public input from the RFI. There is much attention being given to how NICHD can maintain the interest and focus on an important clinical research area that corresponds well with the NICHD mission, "...to ensure that every person is born healthy and wanted..." and "that all children have the chance to achieve their full potential for healthy and productive lives."