

Special Report: 8th Annual Workshop on Malformation and Morphogenesis— August 15-19, 1987, Greenville, South Carolina

Judith G. Hall, M.D.
Associate Editor
Growth, Genetics, and Hormones

Several reports on a variety of congenital defects and inherited syndromes in animals and humans were presented at this workshop. Dr. W. Webster (Sydney, Australia) reported that handling the uterus of the pregnant rat during surgery, or for nonsurgical reasons, can lead to limb defects and central nervous system damage compatible with vascular compromise. This may have clinical implications, although there is no evidence that the same sort of anomalies are seen with manipulation of the human uterus during pregnancy.

Dr. C. Stevens (University of Utah) described a carefully done study of the development of embryonic and fetal palmar and digital creases. Finger creases are well-defined by nine weeks and palmar creases by 12 weeks. These observations are important for timing various effects on limb development.

Dr. S. Clarren (University of Washington) described a carefully controlled experiment assessing "binge drinking" in monkeys. Large doses of alcohol (2.5-4.1 g/kg) given during the first week of pregnancy (and on a weekly basis thereafter) can have a significant impact on the fetal development of the monkey in terms of behavior and cognitive developmental delay measured after birth.

Dr. J. Cordero (Atlanta, Georgia) discussed research that suggested the use of multivitamins before conception may reduce the

risk for neural tube defects and possibly for other congenital anomalies.

Excellent studies by Dr. S. Cassidy (University of Connecticut) on Prader-Willi syndrome and Dr. C. Morris (Phoenix, Arizona) on Williams syndrome provided data that allow much better definition of the natural history of these conditions. The studies also suggest that many features thought to be part of the syndromes—such as obesity in Prader-Willi syndrome and behavior in Williams syndrome—can be modified. Long-term follow-up of patients with Weaver syndrome and Robinow syndrome was discussed by their namesakes.

Several reports suggested the possibility that many disorders with patchy areas of dysplasia (such as the McCune-Albright and Proteus syndromes) may represent somatic mosaicism due to single gene changes and chromosome changes.

Dr. K. Jones (University of California, San Diego) described research in which it was demon-

strated that the supraorbital ridge has a role in inducing the contour of the eyebrow. Individuals with aberrant supraorbital ridges will have aberrant placement of the eyebrows.

Dr. P. Duncan (Downstate Medical Center, Brooklyn, New York) presented an analysis of a family with three generations of Russell-Silver syndrome, a condition that is usually nonfamilial.

Reporting on Joubert syndrome, Dr. D. Flannery (Medical College of Georgia) showed a videotape that visually demonstrated the functional changes that occur in affected patients. These changes are sometimes hard to describe, but recognizing them is essential for an accurate diagnosis. The pattern of respiration in Joubert syndrome is quite striking, with episodic hyperpnea and abnormal eye movements. CAT scans of patients with these breathing and movement patterns may demonstrate the aplasia of the cerebellar vermis associated with Joubert syndrome.

In Future Issues

Osteogenesis Imperfecta: An Update
by Peter Byers, M.D.

Lipodystrophy
by William L. Clarke, M.D.

Medical Complications of the Skeletal Dysplasias
by Judith G. Hall, M.D. and
David L. Rimoin, M.D., Ph.D.

Please send all correspondence to Robert M. Blizzard, M.D.,
Department of Pediatrics, University of Virginia School of Medicine,
Charlottesville, VA 22908.