

Genetic and Environmental Risk Factors for Major Birth Defects Symposium

The four invited speakers, all recognized experts in their field of research, were:

1. Leslie Biesecker, M.D., Senior Investigator, National Human Genome Institute, Bethesda, Maryland; Topic: Polydactyly
2. Juergen Kohlhase, M.D., Ph.D., Professor, University of Freiberg (Germany); Topic: Malformations associated with SALL 1-4 mutations
3. Patrick Brophy, M.D., University of Michigan, Ann Arbor, MI; Topic: Renal Agenesis: Beyond Potter Syndrome
4. Jeanne Manson, Ph.D., MSCE, Research Professor, Children's Hospital of Philadelphia; Topic: Hypospadias

Doctor Biesecker noted that polydactyly is a feature of 119 different disorders, with the specific mutations identified for 39 of those 119. He used the model of the GL13 gene to illustrate how different types of changes were associated with different phenotypes. Professor Kohlhase discussed how haplo-insufficiency in the developmental regulatory gene SALL1 causes the Townes-Brocks Syndrome. He described the wide spectrum of phenotypic features produced by SALL4 mutations. Doctor Brophy reviewed family studies underway to identify members in one of several candidate genes as a cause of familial renal agenesis. He predicted that a cascade of several interrelated genes will be identified as the basis for well-known variations in the associated malformations. Doctor Manson reported on the findings in a detailed search for mutations and environmental factors related to the occurrence of hypospadias. She noted having identified exposure to pesticides in pregnancy as a potential etiologic factor.

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