

Special Report

American Society of Human Genetics Meeting

November 12-15, 1989, Baltimore, Maryland

Judith G. Hall, M.D.

Associate Editor

Growth, Genetics, and Hormones

Among many outstanding symposiums and presentations, some highlights of this meeting included a report by Tsui of the isolation of the cystic fibrosis gene on chromosome 7. According to Tsui, 70% of individuals carrying the cystic fibrosis gene have the same defect (allele). Recent work has attempted to characterize the other 30%. With the isolation of the gene, work on its function and on the pathogenesis of cystic fibrosis becomes the central issue, along with the question of whether newborn screening should be adopted.

Nicholls and co-workers reported several patients with Prader-Willi syndrome who, instead of

having deletions of chromosome 15, had inherited two copies of chromosome 15 from their mothers. Both isodisomy and heterodisomy of maternal chromosome 15 were reported. This suggests strongly that it is the *absence* of the specific locus in the p11-p13 region on chromosome 15 that is responsible for producing the syndrome.

Verlinsky and co-workers presented a new approach to pre-conception prenatal diagnosis, using in vitro fertilization techniques prior to fertilization. After removing the first polar body, they were able to analyze its DNA using the polymerase chain reaction, to see whether it carried an abnormal allele. They were looking for the abnormal allele of the α -1 antitrypsin gene, but almost any other characterized gene could be analyzed in

the same way. If the abnormal allele is in the polar body, the egg will be left with the normal gene; thus prenatal diagnosis can be accomplished prior to fertilization and implantation. The problem with the technique is that (1) crossover occurs with meiosis, and (2) the polar body may be heterozygous.

A large symposium was held on the status of the human genome project. Both the NIH and the Department of Energy are advocating an improvement of techniques for mapping and isolating genes, particularly with regard to technology, management of large amounts of information, and communication between researchers. In addition, a number of issues have arisen relating to the ethics of the research itself and to the ethical uses of the information that is obtained.