

## Special Report:

# Annual Meeting of the American Pediatric Society/Society for Pediatric Research (Genetics Sessions)—April 27-30, 1987, Anaheim, California

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Among the highlights of this meeting was the awarding of the outstanding young investigator prize to Dr. Arthur L. Horwich for his work on the molecular structure of ornithine transcarbamylase. The gene was isolated and sequenced and found to have three sections: a leader peptide necessary for directing the molecule into the mitochondria, a propeptide section requiring removal to activate the enzyme, and the enzyme itself. Using *in vitro* mutagenesis, Dr. Horwich was able to identify the amino acid sequences that led to changes in the various parts of the protein.

Dr. D.S. Rosenblatt reported a new vitamin B<sub>12</sub>-dependent condition in which mild retardation is associated with megaloblastic anemia. The condition is responsive to vitamin B<sub>12</sub> therapy.

Studies of ovarian function in galactosemic patients conducted by Dr. Francine Kaufman and her colleagues indicate that females

who were completely deficient in galactose-1-phosphate uridyl transferase all had ovarian failure. Only those galactosemic patients with partial presence of the enzyme were fertile.

Dr. Ian T. Thomas reported a large number of patients with chromosomal mosaicism that was not necessarily reflected in the karyotypes of the peripheral blood cells. Mental retardation, asymmetry, and striking pigmentary abnormalities, however, were present. Dr. Robert Brent reported that antiserum against yolk sac proteins could have a teratogenic effect and that this effect could not be reversed by vitamin supplementation. Dr. Ira Chasnoff confirmed previous reports that cocaine use by pregnant women is associated with genitourinary tract anomalies in the fetus.

In his paper on Rett's syndrome, Dr. John Moeschlen said that the disorder is much broader in spectrum than had been previously

recognized.

Dr. Grant Mitchell isolated a gene and at least two pseudogenes connected with the ornithine aminotransferase deficiency that may account for gyrate atrophy. Cytogenetic molecular studies of non-disjunction in trisomy 13 syndrome, which were reported by Dr. Terry Hassold, indicate that 60% to 70% of non-disjunction occurs in the first maternal meiotic division, just as it does in Down's syndrome. Molecular studies by Hassold et al demonstrated that, because of lack of crossing over, non-disjunction did not occur. Dr. Carolyn Hadley described experiments in which the gene for phenylketonuria was transferred into hepatocytes via a retrovirus. In his presentation, Dr. Aubrey Milunsky reported that the serum alpha-fetoprotein determination used to screen for chromosome defects was as efficient when used in the first trimester as it was in the second.