

## Special Report: The Western Society for Pediatric Research Meetings— February 3-6, 1987, Carmel, California

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*Growth, Genetics, and Hormones*

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Many excellent papers of interest to endocrinologists and geneticists were presented at the meetings of the Western Research Society. Among them was one by Dr. Ray White, who reported that the order of linkage on chromosome 7 for cystic fibrosis is met-CF-J3111. Dr. White also pointed out that the crossover ratios for different segments of different parts of different chromosomes are different in males and females. There is no consistent pattern, and each arm and arm segment of different chromosomes is being recognized to give markedly different crossover rates.

Dr. Judson Van Wyk delivered the Stanley Wright Memorial Lecture. He discussed peptide growth factors and indicated that there may be fewer peptide growth factors than previously thought, because any given growth factor may act in many different tissues in different ways and under different stimulation. Interestingly, however, a specific peptide growth factor seems to be effective only on neuroectodermal or mesodermal or endodermally derived tissues.

Dr. Larry Shapiro reported that there is clearly a pseudo-autosomal part of the short arms of both the X and Y chromosomes; this portion is shared and has obligatory crossover. Between the

locus on the X chromosome and the centromere there is a steroid sulfatase (STS) locus. On the Y chromosome at the same site there is a pseudo-STS gene, and proximal to that on the Y chromosome is the testes-determining factor. Rarely in human beings is there crossover between the X and Y chromosomes below the pseudoautosomal area; this explains why there are occasional XX males and STS-deficient females.

A memorial symposium in honor of Dr. Joseph St. Geme was established, at which a review of cytomegalic inclusion disease was presented by Dr. Charles Alford. He pointed out that this disease, which leads to extensive teratogenic effects in human beings, is probably now the most serious preventable viral illness in pediatrics. Approximately 35,000 newborns in the United States are affected each year, and at least 20% have significant sequelae.

In his report, Dr. David Rimoin said that the Kniest syndrome, the spondyloepiphyseal dysplasias, and Stickler's syndrome all seem to have linkage to type II collagen. Dr. Hollister reported that Marfan's syndrome seems to be an abnormality of fibrillin. Monoclonal antibodies to fibrillin were used to identify Marfan's patients in a double-blind study. These patients have recognizable fibrillin abnormalities.

Dr. A. Fujimoto described a new autosomal dominant pseudocleft syndrome characterized by a broad nose, colobomas of the eye, and branchial arch involvement.

Dr. Claire Leonard reported several cases of craniosynostosis and facial dysmorphism associated with maternal hyperthyroidism. The thyrotoxic state appeared to trigger early fusion of the cranial sutures.

Dr. Colleen Morris discussed the findings of a survey of 81 patients with Williams' syndrome and defined an evolving natural history that changes from infancy to adulthood.

Dr. Judith Hall reported that gonadal mosaicism appears to be responsible for some autosomal dominant conditions—such as pseudoachondroplasia—seen in siblings with normal parents. As many as 3% of apparent new dominant mutations may actually occur because of gonadal mosaicism for the abnormal gene in one of the parents. That is, during embryologic development of the parent, a somatic mutation occurred, giving rise to some tissues that carry the mutation, but not enough tissues to express the disorder in the parent. However, the gonad or gonads would carry the mutation and therefore the condition can be passed on to more than one child in the family.