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SATURATION MAPPING OF USHER TYPE II REGION ON CHROMOSOME 1q.

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Usher syndrome is characterized by congenital hearing loss, retinitis pigmentosa and differential involvement of vestibular function. It is an autosomal recessive disorder and the frequency of its occurrence is about 3 to 4 out of 100,000 individuals in the United States. The pathogenesis of the disease is obscure and poses a great problem for the treatment. Several studies have shown clinical variability and different types of Usher syndrome have been hypothesized. Recently, the gene for Usher type II (USH2) has been localized to chromosome 1q32-1qter region by linkage with pTHH33 (D1S81) marker (Kimberling et al.,(1990) Genomics 7, 245). The same marker do not show linkage in Usher I families proving genetic heterogeneity and variable clinical expressions are due to non-allelic genes. The mutation causing the disease will not be known until the gene is cloned and characterized. The location of USH2 gene on chromosome 1 has been narrowed down to about 11 cM between flanking markers pTHH33 (D1S81) and CRI-L744 (D1S48). Since there are no markers available in this region to further refine the USH2 region, twenty four new markers have been developed using a flow sorted chromosome 1 library. Three have been shown to be polymorphic. Others are being screened for polymorphism with several different restriction enzymes. Further studies are underway to localize these new markers in the USH2 region by somatic cell hybrid cell lines and in situ hybridization. This will enable us to map the USH2 gene in the range of 1 to 2 cM. Results of polymorphism screening of chromosome 1 flow sorted library and chromosome 1 microdissection library will be presented.