

## Abstract 35

### NEWBORN SCREENING IN FRAGILE X SYNDROME: A PILOT STUDY

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Technological development, new treatments and advocacy efforts are contributing to a push for rapid expansion of newborn screening. However, expanded screening raises a number of concerns and in the case of the *FMR1* mutations has been a topic of considerable discussion since the gene was identified. Because phenotypic features are not evident at birth, Fragile X (FXS) must be discerned through abnormalities in development or behavior. The average age of diagnosis is 30-36 months for full mutation males; consequently, children miss the opportunity to participate in early intervention and parents often have additional children with FXS without knowing reproductive risks. Thus, screening for FXS will allow the identification of a greater number of individuals at risk for the disorder or transmitting the disorder. We have recently begun a pilot study of Newborn Screening in FXS aimed to the determination of allele frequencies in the general population and to the assessment of clinical involvement in the wide variety of fragile X-related phenotypes in the primary and extended families of the newborn probands identified by newborn screening. Using our recently developed PCR method for the identification of premutation and full mutation alleles in the *FMR1* gene, our preliminary data, based on over 2500 newborn blood spots, indicates that the frequency of occurrence of premutation alleles, in males and females, is greater than that previously reported. We have also started to document the degree of clinical involvement in the newborn proband and the extended family members and our experience will be discussed.