

## Abstract 36

### FRAGILE X NEWBORN SCREENING IN THE CLINICAL SETTING

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Newborn screening for fragile X syndrome is being carried out at the UC Davis M.I.N.D. Institute in collaboration with Rush Memorial Hospital, Chicago. The protocol includes approaching patients being seen at the ob/gyn clinic and newborn nursery. Permission is gained to speak with them about the newborn screening project in order to identify newborns with fragile X syndrome. Fragile X syndrome is explained, as is the prevalence. Consented infants identified with the full and pre-mutation will be offered genetic counseling, follow up confirmatory testing and participation in 6 monthly developmental assessments. It is explained that, for infants who are positive, other family members that have not yet been properly identified are at risk to carry the gene mutation. Bloodspot testing according to the protocol developed by Flora Tassone is used as the initial screening tool. Data from October 2008 through June 2009 includes: 1) 409 families offered newborn screening; 2) 307 patients consented to have their newborn screened; 3) 102 patients declined newborn screening. 97 additional patients were approached, but were either non-English speaking (Spanish, Chinese, Hmong, Russian) or underage. 1067 patients were admitted to hospital during the same time period. Five unconsented and 2 consented infants were identified with the pre-mutation. One was seen in clinic for follow up confirmatory blood draw and genetic counseling; the other clinic visit is pending. Several difficulties pertaining to obtaining the consent were identified. These include: 1) inability to cover the newborn nursery on the weekends due to shortage of consenters; 2) need to include Spanish speaking consenters; 3) need to have consenters totally dedicated to this project only; 4) importance of developing a relationship with the parents at the time of consent in order to gain greater reliability of follow up contact should the infant be positive.

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