



Dysmorphology Core Progress Report

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Specific Aim 1

To Insure Consistency and Accuracy of Diagnosis

- Implementation of a standard Protocol to be used at each project site
- Development of a manual to explain and standardize the methods used in performing the physical examination
- Translation of the Dysmorphology Form and Manual into Russian and Ukranian

Specific Aim 1

To Insure Consistency and Accuracy of Diagnosis

- Training of Local Pediatricians
 - 4 local Pediatricians have been trained in Moscow
 - 8 Russian Neonatologists have been trained in Moscow Region
 - 16 Pediatricians, Neonatologists and/or Geneticists have been trained in Ukraine
 - 4 Pediatricians and/or Neurologists have been trained in Rome.

Specific Aim 1

To Insure Consistency and Accuracy of Diagnosis

- Travel to consortium sites to verify diagnosis and update training
 - Prospective study – Moscow Region
 - Jones – 3 trips
 - Robinson – 1 trip
 - Neurobehavioral development of children with FASD in Moscow
 - Jones – 1 trip
 - Prenatal U/S markers of FASD in Ukraine
 - Jones – 3 trips

Specific Aim 1

To Insure Consistency and Accuracy of Diagnosis

- Travel to Consortium sites to verify diagnosis and update training (*continued*)
 - FASD Epidemiology in Italy
 - Hoyme – 1 trip
 - Jones - 1 trip
 - Robinson – 1 trip
 - Del Campo – 1 trip
 - Detecting FASD from Neonatal U/S – Seattle
 - No trips
 - Comparison of 3 Diagnostic Modalities in FASD and Related Disorders in African-Americans
 - Robinson

Specific Aim 1

To Insure Consistency and Accuracy of Diagnosis

- Travel to Consortium sites to verify diagnosis and update training (*continued*)
 - Neurobehavioral Studies in South Africa and the US
 - S. Africa – No trips
 - American Indian Reservations in Northern Plains States
 - Hoyme – 2 trips
 - Robinson 2 trips
 - Neuroimaging studies in Finland
 - Hoyme – 1 trip as part of his sabbatical

Specific Aim 2

To Explore Extent to which Various Degrees of deficient ($\leq 3^{\text{rd}}$ vs $\geq 10^{\text{th}}$ %) anthropometric measurements should be used to enhance specificity of diagnosis without loss of sensitivity

Specific Aim 3

Explore Strategies for diagnosis of FASD in the newborn period or at least during the first year of life

Specific Aim 4

- a. Delineation of the full spectrum of defects.

We have significantly increased the number of children designated “deferred” by broadening the limits of this category-

- ex. 78/256 children seen in Rome were deferred.

- b. Identifying clinical features that are most indicative of future problems in neurobehavioral development.

Specific Aim 5

- Correlation of the clinical diagnosis with that determined by 3-D photography –
 - Hoyme received camera in Helsinki on 11 June 2004
 - Robinson has had camera in Buffalo for last 2 months

Significance

- 1 – Demonstrated our ability to train local pediatricians
- 2 - Demonstrated the effectiveness of ascertaining children with FASD in normal 1st grade classes
- 3 – Developed and field-tested a highly effective physical examination form and manual that have been “field tested”

Plans for Next Year

To be determined by the needs of
the individual consortium sites

Publications

Bakhireva, L. et al.: Effective training of Pediatricians to Diagnose Features of the Fetal Alcohol Syndrome in a Russian Sample. *Alcoholism Clinical and Experimental Research* 2004; suppl. 28 (5):42A