TITLE: **Development of a Visit Assessment Tool to Address Birth Defects and Dysmorphology**

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Introduction: Continuing the observational nature of the National Children's Study (NCS), this formative research project will develop, evaluate, and implement an assessment tool to be used to document and categorize physical features by digital photography and physical assessment. The NCS protocol for the newborn baseline physical assessment currently includes a standardized physical examination and photographs at birth, 6 months and 12 months. The focus of this project is to refine a tool for application to the NCS main study, which can be used to standardize observation and documentation of physical variations and abnormal features in field settings.

Methods: Clinical geneticists and pediatricians from University of Utah (UU), University of Mississippi Medical Center, South Dakota State University, University of California, Irvine (UCI) and San Diego campuses, and Dr Susan Astley, an authority on photographic and training modules at the University of Washington, have joined with NCS collaborators to design the dysmorphology assessment instrument (DAI). In the original planning of the study, the investigators had decided to use the *Elements of Morphology* (http://elementsofmorphology.nih.gov/index.cgi) definitions and illustrations on over 400 phenotypic features as a guide for variations and anomalies needing documentation. Bi-monthly conference calls were followed by two in-person meetings, one at UCI to train in photographic assessment, and the other at the UU to decide on the features not captured by photography that needed inclusion in the DAI physical assessment.

Results: The DAI working group developed the two components of the assessment. The first component is the proposed Photographic Protocol made up of 11 photographic images (9 views of craniofacies and hands and feet), and a 10-second video. The second component is the proposed Physical Assessment list, which was the result of narrowing down the *Elements* list to 25 features that are not captured sufficiently on the Photographic Protocol (see attached instrument and examination, which provides an example of instructions for the training module). An online training module that is comprised of detailed instructions for the Photographic Protocol and Physical Assessment is in development and close to completion (see Figure 1).

Conclusions: Our working group has developed the consensus DAI that includes two components: the Photographic Protocol and the Physical Assessment list. The next steps include finalization of the training module for both components of the DAI; local training of field examiners by the individual centers; pilot study; evaluation of the working DAI and modification of it per piloting; validation studies of DAI; and implementation into NCS.