

Current Research Projects Approved by the Board

NEW:

Assessment of the Sensory Deficits in Individuals with CHARGE Syndrome and Correlation with Genotypic Data. Blake Papsin MD (Principal Investigator), Staff Otolaryngologist Department of Otolaryngology – Head and Neck Surgery University of Toronto, Hospital for Sick Children

There is currently a lack of a detailed analysis of the otolaryngologic senses of hearing, balance, taste, and smell in the body of scientific literature. Furthermore, although studies have been performed to correlate certain phenotypic and genotypic traits, these studies were not conducted from the standpoint of an otolaryngologist, and thus lack the aforementioned detailed sensory analysis. Accordingly we intend to perform such a study with the intention of establishing genotype-phenotype relationships with respect to radiologic, audiometric, vestibular, gustatory, and olfactory parameters.

Such a study does not merely fill a gap in the current body of literature. A careful characterization of the CHARGE phenotype with respect to CHD7 status will allow a better prediction of the potential function and deficits of the newly diagnosed patient. Accordingly, we feel this study will help direct medical care in a proactive fashion. This will hopefully provide for better outcomes and higher overall function in this patient population.

Objective

The aim of this study is to correlate genotypic information with phenotypes and measures of the following: radiographic assessment of the temporal bones and cranial nerves, audiometric data, vestibular testing, olfactory assessment, and gustatory evaluation.

Note: Dr. Papsin and Dr. Alex Osborne will be at the 2011 CHARGE conference to present on Ear Nose and Throat (ENT) issues AND TO ENROLL KIDS IN THIS STUDY.

NEW:

Initial Validation Of A Non-Vocal Pain Assessment Instrument For Individuals with CHARGE Syndrome. Kasee K. Stratton School Psychology Doctoral Candidate Central Michigan University

This study is proposed to evaluate pain non-vocally among individuals with CHARGE syndrome and to provide preliminary validation of an observational, multidimensional, non-vocal pain assessment tool designed specifically for individuals with CHARGE syndrome. This instrument was developed based on the results of Stratton and Hartshorne's (2010) investigation of pain in CHARGE. The purpose of this study is to establish psychometric properties of this instrument and to provide an objective and valid measurement of pain expression in individuals with CHARGE who have limited communication strategies.

A call for participant enrollment was issued via email in February 2011 via List Serve and via email to Foundation members. For additional information, you may contact the researcher. Her e-mail address is: strat1kk@cmich.edu.

Participation in National Disease Research Interchange (NRDI) Database Study:

The NRDI is a non-profit organization funded in part by the NIH to assist in collection and storage of tissue and collaboration with researchers. It is a 30 year old organization and one of their newest initiatives is the rare disease initiative: NRDBA (National Rare Disease Biospecimen Alliance). The purpose of this is to match researchers with specimens from individuals with rare diseases. Specimens can be taken during surgery (e.g. bit of trachea when tracheotomy is performed) or after death (multiple tissues). This program is extremely helpful in collecting and disseminating biospecimens to researchers. NDRI can work with people in the US, Canada and several other countries – they are slowly expanding to more. For the CHARGE Syndrome Foundation to be an active partner, there is a \$1000 yearly fee. Researchers are charged small amounts to receive specimens, depending on the type of specimen, how it has been stored, etc. to the best of our knowledge, donors are not charged anything. NDRI takes care of getting consent, gathering specimens, storing, etc.

The National Disease Research Interchange (NDRI) formed the "Rare Disease Biospecimen Alliance," to extend its services, providing human tissue to Rare Disease scientists. The Alliance

provides a partnership between the CHARGE Syndrome Foundation and NDRI which will target human tissues appropriate for scientific study in your disease area. Working together within this Alliance, we can affect the building of a donor registry, the growth of a researcher database and the coordination of human tissue donations for research from surgical procedures and post mortem. NDRI has customized systems in place to match donors with the appropriate scientists for the placement of valuable human tissues for research. In addition, this collaboration provides for the vital collection of medical history data which accompanies each tissue donated. Please feel free to visit the NDRI website at <http://www.ndriresource.org/>.

Sleep Apnea in CHARGE Syndrome. Dr Kim Blake. IWK Health Center.

There is limited literature concerning the treatment of sleep apnea for children with CHARGE Syndrome. Current research involving general populations of children with sleep apnea shows that adenotonsillectomies improve the children's physiological parameters of sleep. No studies have been published thus far that examine the potential of an adenotonsillectomy to improve sleep apnea symptoms in the CHARGE Syndrome population. Therefore, this research seeks to gather knowledge of the prevalence and symptomatology of sleep apnea in CHARGE Syndrome, as well as to determine the effects of adenotonsillectomy on the symptoms of sleep apnea affecting quality of life of children with CHARGE Syndrome. This research may improve the knowledge base regarding prevalence and symptoms of sleep apnea in the CHARGE Syndrome population.