



CHARGE Syndrome

Bibliography of Resources

Dr. Timothy Hartshorne is a professor of Psychology at Central Michigan University (CMU). In addition, he is the director of the CHARGE Syndrome Lab at CMU. The function of the lab is to investigate the genetic, biological, environmental and individual factors that may be implicated in the etiology of these behaviors, with the aim of better describing the behavior and developing appropriate strategies for intervention and prevention. Listed below are abstracts of some of the research projects that have been conducted through the CHARGE lab. The full text of these articles can be obtained at <http://www.cmich.edu/chsbs/x31482.xml>.

Hartshorne, T.S., Heussler, H.S., Dailor, A.N., Williams, G.L., Papadopoulos, D., and Brandt, K.K. (2009). Sleep Disturbances in Charge Syndrome: Types and Relationships with Behaviour and Caregiver Well-Being. Developmental Medicine and Child Neurology. 51, 143-150.

Children with CHARGE syndrome frequently develop moderate to severe behavior difficulties and are often diagnosed with obsessive-compulsive disorder, attention deficit disorder, Tourette syndrome, and autism. Anecdotal reports have indicated that sleep is also affected. However, the prevalence and types of sleep disturbance have not been identified. This study investigated sleep disturbances in 87 children with CHARGE syndrome, aged 6 to 18 years (mean 11y, SD 3y 8mo). There were 52 males and 35 females represented. Instruments included measures of sleep (Sleep Disturbances Scale for Children [SDSC]), behavior (Developmental Behaviour Checklist [DBC]), and carer well-being (Malaise Inventory). On the SDSC, 57.5% received scores considered significant for sleep disturbances, with disorders of initiating and maintaining sleep, sleep breathing, and sleep-wake transition being the most common. The SDSC was significantly correlated with the DBC ($p=0.010$) and the Malaise Inventory ($p=0.003$). Regression analysis found that both problem behavior and sleep disturbances contributed to the prediction of scores on the Malaise Inventory. Being both deaf and blind ($p=0.001$), experiencing frequent middle-ear infections ($p=0.015$), and starting to walk at an older age ($p=0.007$) were associated with more sleep disturbance. Craniofacial anomalies were not. The study highlights the importance of addressing

the sleep difficulties associated with CHARGE syndrome relating both to airway management and to disorders of initiating sleep.

Reda, N.M. & Hartshorne, T.S. (2008). Attachment, bonding, and parental stress in CHARGE syndrome. Mental Health Aspects of Developmental Disabilities, 11, 10-21.

Parents of 25 children with CHARGE syndrome, ages 12 to 50 months, completed measures of child attachment, parental bonding, and family stress. Twelve children were classified as securely and 13 insecurely attached. The time it took to appear attached and parents to feel bonded were related, as were length of time to appear attached and strength of parental bonding. Visual impairment was related to an insecure attachment as well as to parenting stress. Twelve parents had scores indicating significant stress. Parenting stress was related to problems with bonding, and having a challenging child was related to insecure attachment. Being able to hold the child and a shorter stay in the hospital after birth were related to more secure attachment.

Wachtel, L. E., Harthorne, T.S., & Dailor, A.N. (2007). Psychiatric diagnoses and psychotropic medications in CHARGE syndrome: A pediatric survey. Journal of Developmental and Physical Disabilities, 19, 471-483.

Many children diagnosed with CHARGE syndrome demonstrate behavioral difficulties in addition to visual, hearing and other systemic impairments. Previous research has reported that children with CHARGE have increased rates of self-injury and aggression as well as increased frequency of obsessive compulsive and autism spectrum disorders. This study asked parents to report not only the diagnoses given for their child's behavior problems, but also whether psychotropic medication interventions were prescribed, and which agents were chosen. Results of the study showed that according to parental report, anxiety disorders and pervasive developmental disorders were the most common diagnoses assigned with antidepressant and antipsychotic medications the most frequently prescribed psychopharmacological agents.

Hartshorne, T.S., J. Grialou, t.L. & Russ, A.M. (2007). Executive Function in CHARGE Syndrome. Child Neuropsychology, 13, 333-344.

This study addressed the presence of executive dysfunction in children with CHARGE syndrome, a genetic disorder with multiple physical anomalies and severe challenging behaviors. Ninety-eight children were included in the study. More than half received

clinically significant scores on the Behavior Rating Inventory of Executive Function (BRIEF; Gioia et al., 2000) scales of Shift, Monitor, and the Behavioral Regulation Index, with additional high scores on Inhibit and the Global Executive Composite. Associations were found with the age the child first walked, scores on the Autism Behavior Checklist (ABC; Krug et al., 1993), and being classified as deafblind. Difficulties with making transitions and flexible problem solving, monitoring their work and their effect on others, and acting on impulse, may be related to the behavioral difficulties exhibited by children with CHARGE. Interventions targeting improved self-regulation may help to manage this challenging behavior

Hartshorne, T.S., Hefner, M.A., & Davenport, S.L.H. (2005). Behavior in CHARGE Syndrome: Introduction to the series. American Journal of Medical Genetics, 133A, 228-231.

Challenging behavior in children with CHARGE syndrome has been increasingly a concern of parents, educators, and health professionals. This article introduces the special topic in the American Journal of Medical Genetics on behavior in individuals with CHARGE syndrome. It provides background on CHARGE syndrome, diagnostic criteria, and the relationships of sensory and other physical deficits with both development and behavior. Four themes related to our developing understanding of behavior in CHARGE are described: children with CHARGE have behaviors different from those seen in other syndromes with or without deafblindness. The behavior they display is often very adaptive to their environment and their own disabilities. These behaviors may be partially related to problems with arousal and self-regulation. And, finally, all papers point to behavior as communication, especially within relationships, where it is essential for maximizing intellectual and social outcomes.

Hartshorne, T.S., Grialou, T.L., & Parker, K.R. (2005). Autistic-Like Behavior in CHARGE Syndrome. American Journal of Medical Genetics. 133A, 257-261.

Children with CHARGE syndrome frequently exhibit moderate to severe behavior difficulties, and are often diagnosed with obsessive-compulsive disorder, attention deficit disorder, Tourette syndrome, and autism. Hartshorne and Cypher (2004) surveyed parents of 100 children with CHARGE worldwide and confirmed the prevalence of behaviors that are associated with these disorders. They also found behaviors that could be described as typical of persons who are deafblind. The present study examined whether the autistic like behaviors of children with CHARGE are more similar to those of children who are deafblind, to those of children who are autistic or are unique to

CHARGE. Surveys including the Autism Behavior Checklist (ABC) were mailed to families of 204 children with CHARGE, and 160 usable surveys were returned (78%). Total scores on the ABC for children with CHARGE were significantly different from the norms for those with autism, and those who were deafblind. However, the variance for CHARGE was larger than for the normative groups, and 27.5% of those with CHARGE could be classified as autistic. The pattern of subscale scores for those with CHARGE differed from the other normative groups.

Hartshorne, T.S. & Cypher, A.D. (2004). Challenging behavior in CHARGE syndrome. Mental Health Aspects of Developmental Disabilities. 7(2). 5-7.

Little is known, beyond anecdotal reports, concerning the challenging behaviors of some children with CHARGE Syndrome. 100 respondents from the US and 7 foreign countries completed a web-based survey regarding the behaviors of a person with CHARGE. Included was a medical history and a list of 71 behaviors based on the diagnostic categories most frequently reported anecdotally. Behaviors typical of autistic disorder, attention deficit/hyperactivity disorder, obsessive-compulsive disorder, Tourette syndrome, and deafblindness were characteristic of these children. Those who were deafblind received higher ratings on these challenging behaviors.

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