

CHARGE Syndrome and Autism: Possible Parallels in Challenging Behavior

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Abstract

Special educator and mother of a grown son with CHARGE syndrome reviews data from multiple studies on challenging behaviors in those with CHARGE syndrome in an effort to correlate observed behaviors in her son with possible parallels to autism. This author, special educator, and mother attempts to find parallels between autistic-like characteristics and challenging behaviors in CHARGE syndrome to encourage other special educators and parents of children with CHARGE syndrome to utilize intervention strategies used for students with autism for their students with CHARGE syndrome and similar challenging behaviors. The author includes personal anecdotes regarding her son's behaviors throughout this review.

CHARGE is an acronym for a syndrome of a combination of multiple congenital anomalies. According to Blake, Salem-Hartshorne, Daoud, & Gradstein (2005), the acronym stands for **C**oloboma, **H**ear defects, **A**tresia of the choanae, **R**etardation of growth and development, **G**enital hypoplasia, **E**ar anomalies and deafness and was "coined in 1981 by Pagon and colleagues" (Blake et. al., p. 151).

Several current research studies reflect concern for the emerging evidence about challenging behaviors in children with CHARGE syndrome. Many of these challenging behaviors parallel autistic-like characteristics (Hartshorne, Grialou, & Parker, 2005). As a parent of a grown son with CHARGE syndrome, as well as a special educator, this author was intrigued in reviewing this literature in order to review challenging behaviors observed in her own son she has termed as autistic-like for many years in an effort to help teachers and others relate to his behavior.

Organization and structure are ways he copes. His school binder was always in perfect order in high school. His college backpack is organized and has special places he keeps certain items. His room is always neat and he packs and unpacks drawers and shelves and re-organizes them. He plans for months in advance for trips and talks incessantly about his plans for them. Recently, we told him to wait to talk about camp until two weeks before, but of course, that is impossible for him. Camp is in five months and

yesterday he discussed incessantly which suitcase and why he will take to camp. It's going to be a long five months for me. (L. K. Sheriff)

Autism Spectrum Disorders (ASDs) are characterized by the American Psychological Association the following three feature characteristics: "qualitative impairments in communication and in social interaction, and the presence of repetitive behavior and restricted interests" (Moss & Howlin, 2009). Overlapping of behavior characteristics and ASD features within syndromes such as CHARGE syndrome may aid in better understanding of challenging behaviors for parents and teachers of children with CHARGE syndrome. Perhaps interventions and strategies useful for educating those with autism may be replicated for some students with challenging behaviors and CHARGE syndrome within home and school environments. Knowing the possibility of parallels between autism and CHARGE syndrome may prove advantageous for early intervention strategies for some of these very special education students. As a special educator and mother of a child with CHARGE syndrome, this author would have loved for others to share this connection in early years rather than have discovered some similarities through trial and error experiences.

Method

This article is a review of literature located through the University On-line Library system using key words: autism, CHARGE syndrome, and challenging behavior and dates from 2005-2009. The author considered the last ten years to be current, but in no way can substantiate that other than personal choice as a parent of a grown child with CHARGE syndrome and wanting to study information related to CHARGE syndrome and challenging behavior. As a special educator and conscious of the Individuals with Disabilities Improvement Act of 2004, requiring special educators to teach students with disabilities utilizing evidence-based interventions, the author reviewed literature for scientific-based information. ERIC education and health databases were used and only peer-reviewed journal articles in English were chosen. Several recent 2009 publications were requested through Texas Tech Document Delivery LLIad System. Three of the earlier articles were requested as ancestral articles after the author read about them in some of the later journal articles reviewed.

Literature Review

The first study reviewed was a structured interview study of 36 females & 14 males aged 13-30 years old born with CHARGE syndrome (Blake et al., 2005). The researchers' purpose was to characterize "unique issues" children with CHARGE syndrome have in adolescent years and into adulthood (Blake et. al., 2005, p. 152). The individuals with CHARGE syndrome were recruited for this study through addresses of CHARGE Syndrome Foundation, United Kingdom CHARGE Family Support Group, and participants at the 1999 and 2001 International CHARGE Syndrome Foundation Conferences. At least one parent for each individual with CHARGE syndrome had to agree to be interviewed by the researchers. Structured interviews of 45 minutes

were conducted either in person or by telephone with a parent of each individual with CHARGE syndrome.

Results of the structured interviews showed the following characteristics of CHARGE syndrome found in the participants listed in order of largest to smallest in number: “developmental delay, ear abnormalities, coloboma, cranial nerve dysfunction, cardiovascular malformations, genital hypoplasia, growth deficiency, choanal atresia, orofacial cleft, renal anomalies, tracheoesophageal fistula, spinal anomalies, and thymic/parathyroid hypoplasia” (Blake et. al., 2005, p. 152-153). The average age for walking of these participants was 4.1 years of age. Physicians considered to be regularly visited by participants were in the following specialty fields of medicine: “ophthalmology, otolaryngology, endocrinology, cardiology, psychiatry, orthopedics, neurology, orthodontics, chiropractics, urology, and nephrology” (Blake et. al., 2005, p. 153). Several new medical conditions were reported: “scoliosis (63%), sleep apnea (43%), abdominal colic (40%), retinal detachment (33%), cataracts, migraines (25%), seizures/epilepsy (17%), urinary tract infections (17%), and hypoglycemia” (Blake et. al., 2005, p. 154). Twenty-two (73%) of the participants reported help from psychologists and 18 (62%) took medication for behavior-related issues. All male participants showed late puberty, even two utilizing hormone replacement therapy (HRT). Average puberty for males using HRT was 15.1 years old. Average puberty for girl participants without HRT was 13.3 years and those utilizing HRT was 14.3. Ten of the participants had not reached a level of independence even though they were 13-18 years of age. The average age for toilet training was 5.5 years and all participants had not been diagnosed with CHARGE syndrome until they were older than five.

Recently he stared at a store clerk, raised his voice, and said, “Really? You think I’m a ma’am? Really?”

“He’s been telling me that people think he is a woman. I wish I had listened sooner. I bet that clerk wishes I had, too.” (L. K. Sheriff)

The second study reviewed was of a large group (no number is given) recruited through Metropolitan Atlanta Developmental Disabilities Surveillance Program and the Metropolitan Atlanta Congenital Defect Program to study the combination of Autism Spectrum Disorders (ASD) and “major birth defects” (Fennell, 2009, p. 766). Understanding of etiological pathways leading to autism was the purpose for this study. Participants were between the ages of three and ten years old. This data study revealed that autism “increased twofold” (Fennell, 2009, p. 767) in children with birth defects. A correlation was found in autism related to vision and central nervous system disabilities such as colobomas in CHARGE syndrome and is related to prenatal development time during pregnancy.

The third study in this literature review was conducted to characterize behavioral features of individuals with CHARGE syndrome by comparing behaviors with other syndrome types leading to suggestive educational interventions for students with CHARGE syndrome (Graham, Rosner, Dykens, & Visootsak, 2005). Fourteen males aged 6-21 years of age were recruited for this study through the International CHARGE Syndrome Support Group and university-based geneticists by letter from Cedars-Sinai Medical Center Institutional Review Board. Ten of the 14 boys were legally blind and all were deaf. These 14 males were comparable in age with the following number of boys with other listed syndromes: twenty boys with Down syndrome, 17

with Prader-Willi syndrome, and 16 with Williams syndrome. Parents of the participants completed questionnaires using the Reiss Profile of Fundamental Goals and Motivation Sensitivities, the Achenbach Child Behavior Checklist (CBCL), and the Aberrant Behavior Checklist (ABC).

Results on the Achenbach Child Behavior Checklist (CBCL) showed that the boys with CHARGE syndrome and Prader-Willi syndrome were withdrawn more than those with the other syndromes. Also noted was those with CHARGE syndrome were less anxious than the other three groups of boys. Externalized behaviors were closely related for all the groups of boys. The researchers stated that the boys with CHARGE syndrome “were not at increased risk for aggression or delinquency and appeared at low risk for maladaptive behaviors” (Graham et. al., 2005, p. 242). Scores revealed on the Aberrant Behavior Checklist (ABC) for the boys with CHARGE syndrome showed: “agitation/irritability at 9.0, lethargy/social withdrawal at 6.71, stereotypic behaviors at 5.21, hyperactivity at 8.93, and inappropriate speech at 2.0” (Graham et. al., 2005, p. 242). On the Reiss Personality Profile, the boys with CHARGE syndrome scored lower than the other groups on social contact, showed less attention-seeking behaviors, found keeping order interesting, and scored as equally frustrated as the group of boys with Prader-Willi syndrome. They also showed food interests only second after the boys with Prader-Willi syndrome. These researchers concluded that the boys with CHARGE syndrome have behaviors that resemble autism.

Rules are to be followed. Interrupting other members of the family to correct them is just one of his ways. Just today he told me to “shhhhh” and to not interrupt the doctor. Yes, in front of the doctor. I was embarrassed, too, when he lectured me loudly (he’s deafblind) going to the car about interrupting others. He continued to bring this up and persevere until I fussed back. He cried and rubbed his eyes in frustration with me. He’s nearly 28. (L. K. Sheriff)

The researchers in the fourth study of this review examined sleep problems in individuals with CHARGE syndrome and the correlations between sleep disturbances and challenging behaviors and parental well-being (Hartshorne, Heussler, Dailor, Williams, Papadopoulos, & Brandt, 2009). They also looked for correlations between deafblindness, choanal atresia, and ear infections. The association between walking age and sleep disturbance for those with CHARGE syndrome was also studied. Parent members of the CHARGE Syndrome Foundation responded to questionnaires. Fifty-two males and 35 females with CHARGE syndrome diagnoses given by physicians were the participants in this study via their parents’ questionnaire responses.

The CHARGE syndrome history questionnaire, the Malaise Inventory, the Developmental Behaviour Checklist (DBC, 2nd edition, Parent/Carer version), and the Sleep Disturbance Scale for Children (SDSC) were answered by participating parents (Hartshorne et. al., 2009). Results concluded that challenging behaviors follow sleep disorders. The researchers noted several problematic sleep scenarios. The foremost sleep issues for participants with CHARGE syndrome are in falling asleep and staying asleep. Breathing during sleep issues and waking-up transition times proved significant within the areas of self-absorbed challenging behavior. Over half of the participants show problematic sleep issues within clinical score range. The participants with deafblindness scored higher on the SDSC test and there was no correlation between the disabilities of choanal atresia and cleft palate to sleep problems. There was a

correlation between sleep issues and ear infections in participants. Scores on the Malaise Inventory corresponded to the scores on the Developmental Behaviour Checklist (DBC-P) for parents revealing that sleep issues in children with CHARGE result in sleep issues and problems for their parents.

Many nights I have sat and listened to him breathe as he slept, not able to leave him and wondering if his struggles breathing are due to colds in tandem with choanal atresia. Lately, he tosses and turns due to his crooked spine and complains of back pain keeping him awake. Pain medication is not optimal due to his anticoagulant medication for his aortic valve replacement. Having him cranky is the tradeoff for lack of sleep. Having me cranky is the tradeoff for living with him. Cranky is okay with me for both of us. The alternative isn't. We'll catch a nap when we can. (L. K. Sheriff)

The fifth research study in this review examines executive function in individuals with CHARGE syndrome to determine if correlations to challenging behaviors occur in participants (Hartshorne, Nicholas, Grialou, & Russ, 2007). Parents of 98 children with CHARGE syndrome, ages five to 18, were recruited through a mailing list from the CHARGE Syndrome Foundation. Parents completed the following questionnaires: the CHARGE history questionnaire, the Behavior Rating Inventory of Executive Function (BRIEF) and the Autism Behavior Checklist (ABC). Ninety-eight participant parents returned their questionnaires and 92.5% of them were completed by mothers. The average age child participant was 10.63.

Results revealed “more than 50% of the children in this study scored in significant range on Shift, Monitor, and Behavioral Regulation Index” (Hartshorne et. al., 2007). On the Autism Behavior Checklist the participants scored higher than norms for individuals with deafblindness without CHARGE syndrome. Those with CHARGE syndrome and deafblindness in this study scored higher on the Behavior Rating Inventory of Executive Function (BRIEF).

Included sixth in this literature review is a study performed to “objectify autism-specific behaviors in people with deafblindness” (Hoevenaars-van den Boom, Antonissen, Knoors, & Vervloed, 2009, p. 550) and aid in appropriate diagnoses of autism in individuals with deafblindness. Ten participants with deafblindness and intellectual disabilities from institutions were the participants, five with autism and five without. Two male participants had CHARGE syndrome and autism. All ten had cognition less than 24 months and the average age of those with autism and deafblindness was 18 years of age. The average age of the five without autism was 15 years. The majority with autism were males and the majority without were females. Semi-structured observations took place at the participants' institutions and lasted from 30 to 60 minutes. Autism and deafblind specialists on this research team developed the Observation of Characteristics of Autism in Persons with Deafblindness (O-ADB). The O-ADB was based on the Autism Diagnostic Observation Schedule of Lord, the Autism Screening Instrument for Educational Planning of Krug, the Autism Diagnostic Interview-Revised of Le Couteur and the Van Dijk Approach to Assessment. Combined together these instruments formed a “semi-standardized observation instrument” (Hoevenaars-van den Boom et. al., 2009, p. 551).

Revealed in the study were individuals with deafblindness and autism were less open, less apt to initiate, and respond to contact. They displayed stereotypies, had change-coping issues and less variety in their behavior. They tended to show lack of problem solving and lack of asking for help to problem solve. Autism was significant on the O-ADB scale, but gender was of

no significance. The two males with CHARGE syndrome showed some behaviors that possibly are distinctly different from those with deafblindness only.

Lately, he eats in the student union. It's always Chik-fil-a sandwiches with an extra pickle packet. He dresses every day in college logo shirts and shops for more of them each month when he has extra cash to spend. He has enough SFASU shirts for three guys to wear. They have become a daily uniform. He asks to go to Chik-fil-a anytime we are out shopping. I even bought frozen chicken patties so he could make his own, but they don't taste the same, I am told....funny, since his sense of taste is barely present if present at all. (L. K. Sheriff)

The seventh study in this literature review series was a systematic search of seven total articles (Moss & Howlin, 2009). This study was an attempt to discover correlations between certain genetic syndromes and Autism Spectrum Disorder (ASD). Also studied was the inclusion of intellectual disabilities along with the syndromes and ASD and to discover whether there could be "clinical implications" leading to diagnoses and interventions for genetic syndromes with ASD. The researchers systematically reviewed papers with ASD and syndromes in the title. Three CHARGE syndrome studies were located and included 191 individuals with CHARGE syndrome.

The researchers determined that "autistic-like behaviors have been described" and specifically that "Hartshorne et. al. (2005) has found variability in the severity of autistic symptomatology in comparison with individuals with ASD, but the results indicated that the presence of autism characteristics in CHARGE could not be wholly accounted for by the visual and hearing impairments typically associated with the syndrome" (Moss & Howlin, 2009, p. 862).

Eighth in this literature review of studies is to answer what characteristics differ for children with CHARGE syndrome as compared to the characteristics of those who are deafblind and those with autism (Hartshorne, Grialou, & Parker, 2005). Participants were parents of 160 individuals with CHARGE syndrome. Two different medical history forms were completed by the parent participants along with the ABC portion of the Autism Screening Instrument for Educational Planning, Second Edition.

Study results indicated that age of a child with CHARGE syndrome did not correlate with ABC scores and scores did not increase or decrease with age. One age category, the three and four year olds, had the highest average score on the ABC. The participants in the 11-14 year old category had scores fall below those for individuals with deafblindness. Concluded was that 27.5% of those in this study could have diagnoses of autism. The ABC scores reveal that those individuals with CHARGE fall lower than those with autism and higher than those with deafblindness. The scores for body object and sensory use were in the middle of autism and deafblind averages. Language was fairly equal to that of individuals' with deafblindness, but in the areas of social and self-help skills, participants scored similar to the autism group studied. The participants who walked when they were older scored higher in the area of autism.

He was top heavy and would topple over, so I held his hands and walked behind him. He learned to walk alone at age three, though he pulled to stand beside a table or bed at age 18 months. He would roll to a chair or table, push and pull himself to his knees, then hold

on with his hands and pull himself up to stand on one leg, then pull up more until he was standing and holding on. If he let go of what he was holding on to, he would fall backwards or sideways as a whole. He never learned to crawl or to fall onto his bottom in a seated fashion. (L. K Sheriff)

The ninth study reviewed was conducted to discover “possible associations between behavior characteristics and medical complications” (Vervloed, Hoevenaars-van den Boom, Knoors, van Ravenswaaij, & Admiraal, 2006, p. 851). A second focus of the study was to determine whether individuals with CHARGE syndrome can be distinguished by their medical issues and how many behavioral issues they exhibit. Fourteen males and 13 females were recruited from all over the Netherlands for this study. Nine were located through the CHARGE parent federation, eight from a residential center for those with deafness or deafblindness, and the remaining from the Otorhinolaryngology of Radboud University Mijmegen Medical Centre.

Medical conditions, hospital stays, and amount and etiologies of participants’ medical conditions were studied. Three questionnaires were filled out prior to a home visit, which lasted 75-195 minutes. Structured interviews took place in the home where medical and psychological reports were also analyzed. Several instruments made of the total of questionnaire materials. These materials included: A 71 question survey, the Questionnaire destine’aux families d’enfants ayant le syndrome de CHARGE, the Temperamentsschaal Voor Zwakzinnigen (TVZ) temperament scale, Child Behavior Checklist (CBCL), and the communication scale of the Vineland Adaptive Behavior Scales (VABS).

Results indicated no correlation between number of medical issues and age of participant to behavior issues. There were negative relationships between hospitalization, cardiac defects and surgery, and with brain abnormalities. Presence of colobomas did not correlate with behavior issues. Developmental delay correlated with deafblindness. The participants with deafblindness showed lower communication scores. Positive behaviors correlated with heart surgery and tube feeding. “the participants with heart surgery especially, had less behavior problems” (Vervloed et. al., 2006, pg. 851). This particular study did not find correlation significance between medical conditions and autism, but the researchers state that visual disability “alone cannot explain the high prevalence of autism in CHARGE syndrome” (Vervloed et. al., 2006, p. 860).

He began to gain weight when I blended real food with milk I purchased from a local goat farmer and put into his feeding tube. We played games as I fed him through his tube. I kept my face close to him and made silly sounds. I dipped a pacifier in honey and rubbed it to his lips as he ate, I gave him suckers and rubbed his tongue with them as he fed through his tube. He responded positively to the attention and loved my closeness during feedings. I could tell because he would laugh and kick his legs in anticipation of the stimulating games we played. (L. K Sheriff)

The tenth study examined was conducted to discover what behavioral issues lead to “single or multiple psychiatric diagnoses” (Wachtel, Hartshorne, Dailor, 2007, p. 473), what medications are prescribed for specific psychiatric symptoms, and whether individuals with CHARGE syndrome are treated with single or multiple medications. Parent participants of 87 children with CHARGE syndrome from 27 different states were sent questionnaires. Fifty-nine percent of child participants were male, 88.5% were Caucasian, and 89.7% of the questionnaires

were completed by mothers. The CHARGE History questionnaire and a Developmental Behaviour Checklist (DBC, Second Edition) were sent to previous parents who participated in previous studies with Dr. Hartshorne were recruited for this study. Two open-ended questions were asked on the questionnaire. They were: whether the child participant had a diagnosis for challenging behavior such as a diagnosis for autism and what supplements or medications their child takes regularly.

Ten participants had anxiety disorders in the form of obsessive-compulsive disorder (OCD) and took antidepressants (mainly SSRIs). Data showed antihypertensive medications and stimulants were prescribed for Attention Deficit Hyperactive Disorder (ADHD) in CHARGE participants, and antipsychotics were dispensed for those with Pervasive Developmental Disorder (PDD). Whether these medications are successful in helping the participants was not discovered in this study.

His backpack is packed with each item in its place. He dusts his room without being told and sits in front of Astros baseball games and scores the plays in scorebooks. He works at puzzles for hours on end to complete them. He leaves an hour before he needs to just to make sure he is on time. Forget trying to reason with him that you will get him somewhere on time; you might as well take him early and save yourself the grief. Just today he told me that procrastination is the devil's workshop. Wonder where he heard that? Not from me, for sure! (L. K. Sheriff)

The eleventh and final study in this literature review of challenging behavior and children with CHARGE syndrome was to test a previous hypothesis made by Reda & Hartshorne (2008) that rearing a child diagnosed with CHARGE syndrome is related to higher stress levels (Wulffaert, Scholte, Dijkxhoorn, Bergman, van Ravenswaaij-Arts, & van Bercklelaer-Onnes, 2009). The study also was to test whether behavioral and physical characteristics of individuals with CHARGE syndrome are related to stress in their parents. Correlations between syndrome-specific features and parenting stress were also explored.

Sixteen boys and six girls with CHARGE syndrome and their parents were solicited by mail from a list of parents in the Dutch CHARGE Parent Support Group. Ages of the children were 1.7-22.2 years of age. Parent participants were sent several questionnaires to complete for this study. Included materials were: the Nijmegen parenting Stress Index-Short (NPSI-S) which is an adapted version of the Parenting Stress Index by Abidin, the Vineland Screener 0-12 years (VS 0-12) for adaptive functioning level, the Dutch version of the Developmental Behavior Checklist-Primary Carer (DBC-P parent rating scale, and expressive communication for the child was determined by the DBC-P questionnaire.

Parenting stress was reported by two-thirds (59%) of the parents in this study. Challenging behaviors in "depression, autism, self-absorbed and disruptive behavior correlated positively with parenting stress" (Wulffaert et. al., 2009, p. 301). Aloofness, sleep disturbances, stubbornness, disobedience, having tantrums, being over-excited and noisy, irritable, having attention difficulties and impatient behaviors were in more than half of the children studies. Impatience was present in 86% of participants.

Limitations

The author of this literature review observes and records that several of the studies involve questionnaires to parent groups of children with CHARGE syndrome recruited through the CHARGE Syndrome Foundation. This may mean many of the questionnaires for different studies may involve the same participants.

Conclusion

The studies in this literature review reflect an emerging cluster of studies paralleling challenging behaviors in children with CHARGE syndrome with some of the same challenging behaviors recognized within Autism Spectrum Disorder (ASD). Parenting stress is also reviewed in several of the studies included in this collection of professional literature. Since American and European researchers have used different assessment instruments, there is definitely evidence supporting autistic-like behaviors in children born with CHARGE syndrome may differ from autistic-like behaviors in other children with multiple disabilities.

Future Implications

Inclusive education settings may be viable options for more students with CHARGE syndrome if scientific-based information paralleling autism and CHARGE syndrome could be recognized and used to address individual students with challenging behaviors. Strategies for students with autism could be paralleled and implemented early in life for many special education students with CHARGE syndrome leading to earlier introductions to inclusive settings in schools for those with CHARGE syndrome. Earlier introduction to inclusive classroom settings could lead to better educational outcomes for special education students with CHARGE syndrome.

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