



Professional Day Poster

CHARGE Syndrome Checklist: Health Supervision Across the Lifespan

Angela Arra-Robar, RN MSN

Presenter Information

Angela Arra-Robar is a Clinical Nurse Specialist with children with medical complexities at the IWK Health Centre in Halifax, Nova Scotia. Angela is a graduate of the University of Western Sydney (Australia) and Canyon College (USA) where she completed her Masters of Science in Nursing in 2006. Over the past twenty-one years, Angela has held a variety of nursing positions in both Canada and the United States. Most of her clinical experience has been in the specialty of medical-surgical nursing in both adult and pediatric settings. She has worked as a staff nurse, clinical instructor and nurse educator prior to her current role as a CNS.

Angela has worked with children with CHARGE syndrome in Atlantic Canada for the past 13 years. Together with Dr. Kim Blake, they run a monthly clinic specifically for children and youth with CHARGE in order to provide continuity in care and support youth and their families to be able to self-manage their healthcare. Angela's passion for teaching is evident through her collaboration with staff, patients and their families to share knowledge, build capacity and promote a truly family-centred care environment.

Presentation Abstract

A comprehensive approach to health screening and management for individuals with CHARGE syndrome is essential. We developed a checklist organized by body system and age to guide the healthcare provider in their approach to care. The checklist was evaluated using a modified Delphi method to develop a final consensus. We anticipate that these recommendations will promote improvements in care by preventing missed diagnoses, allowing for anticipatory counseling, and facilitating early referral for interventions and treatments.

CHARGE SYNDROME CHECKLIST: HEALTH SUPERVISION ACROSS THE LIFESPAN (FROM HEAD TO TOE)

**Shaded boxes indicate key assessment points*

		INFANCY (0-2 years)	CHILDHOOD (3-11 years)	ADOLESCENCE (12-17 years)	ADULTHOOD (18+ years)
GENETICS	Clinical diagnosis (Blake et al. or Verloes or Hale et al. criteria)				
	Genetic testing – Genetics consult (CHD7 analysis, array CGH)				
	Genetic counselling				
NEUROLOGY	CNS malformations/hypoplasia olfactory bulb/temporal bone (semi-circular canal) malformations – requires MRI/CT				
	Seizures – more common at older ages – consider EEG				
	Cranial nerve problems – monitor for absent sense of smell, facial nerve palsy, sensorineural hearing loss, vertigo, swallowing problems				
EYES, EARS, NOSE AND THROAT	Coloboma, risk of retinal detachment - Ophthalmology consult (dilated eye exam in infancy, vision assessments)				
	Corneal exposure – lubricating eye drops				
	Photophobia – tinted glasses, sunhat				
	Choanal atresia/cleft palate/tracheoesophageal fistula - ENT/Plastics consult				
	Audiometry and tympanometry, monitor for recurrent ear infections				
	Adaptive services for individuals with deafness/blindness				
	Cochlear implant assessment if applicable				
	Obstructive sleep apnea – monitor for tonsil/adenoid hypertrophy				
	Excessive secretions – consider Botox, medication				
	Dental issues – consider cleaning under anaesthetic				
CARDIOLOGY RESPIROLOGY	Cardiac malformations common – major/minor defects, vascular ring or arrhythmias possible (echocardiogram, chest x-ray, ECG) - Cardiology consult				
	Sinusitis, pneumonia, asthma - monitor				
	Anesthesia risk (difficult intubations/post-op airway obstruction/aspiration) – extensive pre-operative assessment, combine surgical procedures				
GASTROENTEROLOGY GENITOURINARY	Gastroesophageal reflux – Gastroenterology consult – consider motility agents with proton pump inhibitor				
	Poor suck/chew/swallow - feeding team assessment/intervention				
	Aspiration risk, tracheoesophageal fistula – swallowing studies				
	May need supplemental feeds – frequently requires gastrostomy tube or gastrojejunostomy tube				
	Constipation – consider Senna glycoside with polyethylene glycol				
	Renal anomalies – abdominal u/s +/- VCUG, blood pressure monitoring				
ENDOCRINOLOGY	Hypogonadotropic hypogonadism – LH, FSH by 3 months				
	Genital hypoplasia (if undescended testes - consider orchidopexy)				
	Delayed puberty – Endocrinology consult - gonadotropin levels, HRT				
	Osteoporosis – DEXA scan				
	Poor growth – Endocrinology consult – GH stimulation test, GH therapy				
	Obesity - monitor				
	Fertility and contraception - discuss				
IMMUNE SYSTEM	Note presence of thymus at open heart surgery				
	Routine immunizations/antibody titres to immunizations in adolescence				
	Recurrent infections – Immunology consult				
MSK	Scoliosis/kyphosis- monitor				
	Mobility (affected by ataxia, hypotonia) - evaluate				
PSYCHOLOGY DEVELOPMENTAL	Assess gross and fine motor skills – Occupational Therapy, Physiotherapy				
	Communication, language, writing abilities – Speech Language Therapy				
	Consider deaf-blind consultant				
	Prepare for transitions to school, situations, places, systems				
	Psychoeducational assessment, Individualized Education Plan				
	Sleep disturbances – consider melatonin				
	Behavior management – self-regulation, impulse control, anxiety, obsessions, compulsions, anger				
	Toileting skills - support				
	Life skills/adaptive behaviour/social skills/social play				
	Address sexuality				
	Family stress – offer supports and resources				
	Medical self-management – work on managing medications, understanding conditions, seeing healthcare provider independently				

CHARGE SYNDROME CHECKLIST: HEALTH SUPERVISION ACROSS THE LIFESPAN (FROM HEAD TO TOE)

Abbreviations Used in Checklist

CGH - comparative genomic hybridization
CNS - central nervous system
CT - computed tomography
DEXA scan - dual energy X-ray absorptiometry
EEG - electroencephalogram
ENT - ear, nose and throat

FSH - follicle stimulating hormone
GH - growth hormone
HRT - hormone replacement therapy
LH - luteinizing hormone
MRI - magnetic resonance imaging
MSK - musculoskeletal
U/S - ultrasound
VCUG - voiding cystourethrogram

Resources

- ☐ The CHARGE Syndrome Foundation (<http://chargesyndrome.org/about-charge.asp>)
- ☐ The CHARGE Informational Pack for Practitioners (SENSE UK) (<https://www.sense.org.uk/content/charge-information-pack-practitioners>)
- ☐ Book - CHARGE Syndrome (Genetics and Communication Disorders), 1st ed. Hartshorne TS, Hefner M, Davenport S, Thelin J. 2011
- ☐ OMIM Entry #214800 CHARGE Syndrome (<http://www.omim.org/entry/214800>)
- ☐ CHARGE Syndrome International Conference
- ☐ CHARGE Syndrome Listserv
- ☐ CHARGE Syndrome Facebook Group
- ☐ Perkins School for the Blind e-learning (<http://www.perkinselearning.org/videos>)
- ☐ Deafblind International (<http://www.deafblindinternational.org/index.htm>)
- ☐ Open hands, open access: deaf-blind intervener learning modules (<http://moodle.nationaldb.org>)

Key General References

1. Blake K, Prasad C. 2006. CHARGE syndrome. Orphanet J Rare Dis 1: 34
2. Brown D. 2005. CHARGE syndrome “behaviors”: challenges or adaptations? Am J Med Genet Part A 133A: 268-272
3. Hsu P, Ma A, Wilson M, Williams G, Curotta J, Munns CF, Mehr S. 2014. CHARGE syndrome: a review. J Pediatr Child Health 50: 504-511



Professional Day Poster

What do I have today? Calendar Systems and Sensory Impairments

Shanti Brown and Timothy S. Hartshorne, Ph.D.

Presenter Information

Shanti Brown is a second year Ph.D. student in a School Psychology program. She is a part of the CHARGE Syndrome research at Central Michigan University. Her current research project is investigating the use of calendar systems as an anxiety intervention for children with multi-sensory impairments.

Presentation Abstract

A calendar system is a tool used to refer to and describe an activity by using objects, pictures, and/or other symbols to communicate a sequential process. This presentation will provide descriptions of several different types of calendar systems, including what they look like and how they are used. Calendar systems have been shown to benefit those who have sensory impairments in many ways by increasing communication, independence, security, and social development.



Professional Day Poster

Anxiety in CHARGE Syndrome

Shanti Brown and Timothy S. Hartshorne, Ph.D.

Presenter Information

Shanti Brown is a second year Ph.D. student in a School Psychology program. She is a part of the CHARGE Syndrome research at Central Michigan University. Her current research project is investigating the use of calendar systems as an anxiety intervention for children with multi-sensory impairments.

Presentation Abstract

Anxiety has been found to be a growing concern regarding individuals with CHARGE. This presentation is a synthesis of the current research on anxiety and related behaviors in CHARGE as well as its prevalence in and effect on the population as a whole. The current debate on Obsessive-Compulsive Disorder in individuals with CHARGE will be discussed, as well as the use of interventions aimed at lowering anxiety.



Professional Day Poster

Growth in CHARGE syndrome

Dieuwerke Dijk, MD

Presenter Information

Dieuwerke Dijk is a resident in training to become a physician for people with intellectual disability. Apart from her clinical work at a health care centre for people with intellectual disability, she participates in the CHARGE research at the University Medical Centre Groningen. Scope of her research is growth in CHARGE syndrome.

Presentation Abstract

Growth is a strong indicator of the well-being of children. Although growth retardation is one of the main characteristics of CHARGE syndrome, at present little is known about normal growth in children with CHARGE syndrome. We want to find out more about normal growth in CHARGE syndrome by making growth charts and assessing body proportions in children with CHARGE syndrome.



Professional Day Poster

Dissecting the causes of frontal cortex abnormalities in a mouse model for CHARGE syndrome

Alex P A Donovan

Presenter Information

Alex Donovan is a first-year PhD student at King's college London studying the developmental origins of executive functioning deficits in CHARGE syndrome. Alex has a neuroscience bachelor's degree from KCL, and is now working under Dr. Albert Basson, focusing on the role of the chromatin remodeling factor CHD7 in the development of the frontal cortex, and the consequence of its disruption in CHARGE syndrome.

Presentation Abstract

Many individuals with CHARGE syndrome have been reported to have deficits in executive functioning, often associated with a diagnosis of an autism spectrum disorder. The neurodevelopmental and neuroanatomical underpinnings of these deficits are not yet known. However, functional studies have identified the prefrontal cortex as the primary brain region governing executive functions. I report studies on a *Chd7*^{+/-} mouse model to investigate the impact of *Chd7* heterozygosity on the development and function of the frontal cortex.



Professional Day Poster

CHARGE Syndrome and the IEP: What do parents think?

Whitney Driskell, Liddie McPhail, Hailey Ripple, Tierra Kilbert, Kasee Stratton

Presenter Information

Whitney Driskell is a graduate student at Mississippi State University in the School Psychology Program. Whitney has been a member of the Bulldog CHARGE Syndrome Research Lab at Mississippi State University since January of 2015. Under the advisement of Dr. Kasee Stratton, Whitney serves on the IEP research team. Whitney plans to attend internship and graduate from the school psychology program where she hopes to gain employment in a public school setting and become an advocate for individuals with low incidence disabilities.

Presentation Abstract

Limited research has been conducted on educational accommodations in CHARGE. Additionally, parents have frequently expressed frustration with navigating their child's Individualized Education Plan (IEP). This study aimed to identify parental 1) perceptions of the IEP process, 2) understanding of their child's goals and accommodations and 3) satisfaction with the IEP and the IEP team. The researchers will present their research and how this investigation will advance future educational research.



Professional Day Poster

CHARGE Syndrome in Brazil

Debora Ferreira and Timothy S. Hartshorne, Ph.D.

Presenter Information

Debora Ferreira is a senior at Central Michigan University who work in the CHARGE Syndrome Research Lab. As a dual citizen with Brazil, she is interested in how to support individuals with CHARGE in that country.

Presentation Abstract

This poster is based on information from parents and professionals in Brazil who have a connection to someone with CHARGE syndrome. They are working to learn more and make others aware of this diverse syndrome. Not only do they want to raise awareness and support each other, they are working towards getting more professionals to understand CHARGE so they can better the lives of the ones they know with this syndrome.



Professional Day Poster

You Asked, We Listened: What's missing on the IEP for students with CHARGE?

Matthew Ferrigno

Presenter Information

Matthew Ferrigno is a first year School Psychology doctoral student at Mississippi State University. He joined the Bulldog CHARGE Syndrome Research Lab at Mississippi State University in the fall of 2016. Matthew has been assisting a research team examining individualized education plans (IEP) for students with CHARGE. Due to the lack of specific accommodations and outlines for IEP's, Matthew plans to promote a better understanding of what needs to be placed on an IEP.

Presentation Abstract

Parents often face many complications obtaining special education services and developing IEPs for their children with CHARGE. The Texas Deafblind Outreach has developed a set of IEP quality indicators. Quality indicators not only serve as a guide for those developing IEPs, but they also increase the likelihood of appropriate provision of services. This study will review how IEPs for students with CHARGE compare to the quality indicators provided by the Texas Deafblind Outreach.



Professional Day Poster

Radiological Imaging Findings in CHARGE Syndrome

**Lauren Fogarty, MD Candidate 2020,
Kim Blake MD, Pierre Schmit MD**

Presenter Information

Lauren is a first year medical student at Dalhousie University. She is working with Dr. Kim Blake and Dr. Pierre Schmit researching radiological imaging in children with CHARGE syndrome at the IWK Health Centre in Halifax, Nova Scotia. Prior to beginning her medical degree, Lauren completed her MSc studying molecular signaling throughout neural development.

Presentation Abstract

Individuals with CHARGE Syndrome often have complex medical needs requiring multiple radiological investigations from birth through to adulthood. CHARGE characteristics affect multiple organ systems and can vary significantly from one individual to another. Here, we have reviewed the literature and outlined common imaging findings in children with CHARGE syndrome integrating cases from the IWK Health Centre to illustrate these findings.



Professional Day Poster

Variables that Impact Walking in Individuals with CHARGE Syndrome

Dr. Elizabeth (Beth) Foster, CAPE

Presenter Information

Dr. Beth Foster is a Certified Adapted Physical Educator. She worked as an Elementary Adapted Physical Educator in Pennsylvania for 6 years then was an APE consultant in Texas for 4 years. During her time teaching in Pennsylvania, Beth was named the 2012 adapted physical education teacher of the year. Beth has been involved in Camp Abilities, a developmental sports camp for individuals who are blind, visually impaired, or deafblind for the past 12 years. She is currently the assistant director for Camp Abilities in Pennsylvania and Texas. From her experience at Camp Abilities, she developed a passion and determination to be knowledgeable within the field of deafblindness. Beth has presented on various topics about deafblindness across the U.S. She completed intervener training at the Minnesota Deafblind Project. Beth has been involved with the Texas Chargers for the past 3 years providing physical activities for individuals with CHARGE Syndrome at their annual retreat and has directed the Camp Abilities at the past two International CHARGE Conferences. Beth received her PhD in May 2016 by completing research related to children with CHARGE Syndrome and walking. Currently Dr. Foster is an assistant professor at Cal Poly Pomona in California in adapted physical education.

Presentation Abstract

Children with CHARGE syndrome are often delayed in the attainment of independently walking. The purpose of this session is to provide information about variables that impact walking and the benefits of walking in individuals with CHARGE syndrome. The session provides strategies to develop meaningful and accessible movement activities to enhance walking and provide children with movement confidence. Activities based on best practices and parent input will be presented. Participants leave with strategies that can be implemented when they get home.



Professional Day Poster

The Sensory Base of Unusual Behaviors in Children with CHARGE

Aaron Hartshorne and Timothy S. Hartshorne, Ph.D.

Presenter Information

Aaron Hartshorne is a first year student at Central Michigan University where he is a member of the CHARGE Syndrome Research Lab. He will be transferring to Alma College in the Fall. He is the sibling of a young adult with CHARGE syndrome.

Presentation Abstract

Children with CHARGE syndrome display an array of behaviors that some may consider problematic. However, many times these behaviors are sensory based in nature, and serve to help the child self-regulate and orient his or her body in space. This poster suggests the most probable sensory function of these behaviors.



Professional Day Poster

The Behavior Triangle for CHARGE Syndrome

Timothy S. Hartshorne, Ph.D.

Presenter Information

Tim Hartshorne is a professor of psychology, specialized in school psychology, at Central Michigan University. He is the grant holder for DeafBlind Central: Michigan's Training and Resource Project, which provides support to children who are deafblind in Michigan. Much of his work is influenced and motivated by his son Jacob, who was born in 1989 with CHARGE syndrome. Tim's particular interests include understanding the challenging behavior exhibited by many individuals with deafblindness, CHARGE, and related syndromes, and also how severe disability impacts the family. He is the lead developer of a deafblind intervener training module on behavior for the National Center on Deaf-Blindness. He has been awarded the Star in CHARGE by the CHARGE Syndrome Foundation. His research was recognized in 2009 with the Central Michigan University President's Award for Outstanding Research. He is a frequent presenter on CHARGE and deafblindness.

Presentation Abstract


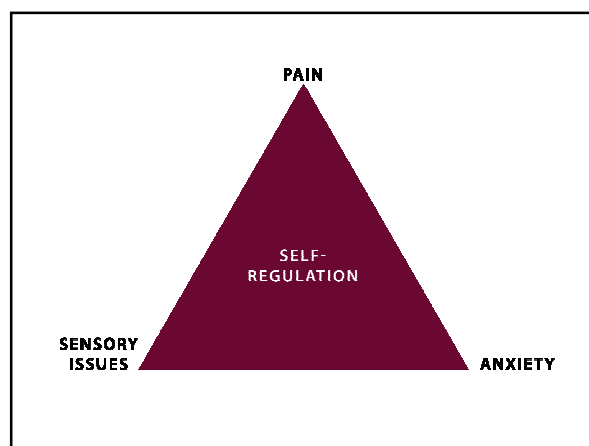
Having considered issues around challenging behavior in individuals with CHARGE for many years, I have come to the conclusion that the behavior has three primary sources: pain, sensory issues, and anxiety. This presentation will address what we know about each of these, and then discuss the development of self-regulation skills as a prime intervention.

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Understanding Behavior in CHARGE Syndrome

Timothy S. Hartshorne, Ph.D.
Central Michigan University

Sources of Pain in CHARGE

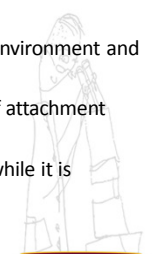
- Otitis Media¹
- Sinus infections
- Migraines¹
- Abdominal Migraines^{1,2}
- Gastroesophageal reflux disease (GERD)²
- Gas
- Constipation^{1,2}
- Muscle pain²
- Tactile defensiveness
- Stoma pain
- Oral pain/teeth
- Surgery¹



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Why pain is so important


- Can affect normal brain and neurological development
- Can affect sleep
- Can interfere with exploration of the environment and learning
- Can interfere with the development of attachment and trust
- Once tolerated may be ignored even while it is affecting health and behavior
- Is communicated through behavior



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Sensory Issues

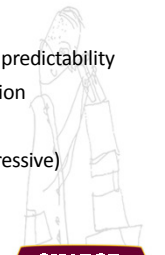
- Hearing – sensorineural hearing loss
- Vision – coloboma
- Smell – anosmia
- Taste – prefer strong tastes
- Tactile – defensiveness
- Vestibular – balance issues
- Proprioceptive – awkwardness
- The loss of each sensory system multiplies the impact on the child and the child's development



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Sensory Processing Disorder

- Missing, partial, distorted, fragmented information
- Over-sensitivity or Under-sensitivity
- Processing time may be longer
- Confusion & the need for consistency & predictability
- Idiosyncratic behaviors & misinterpretation
- Movement and postural differences
- Communication issues (receptive & expressive)
- Developmental delay
- Fatigue



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Anxiety

- “A feeling of worry, nervousness, or unease, typically about an imminent event or something with an uncertain outcome.”



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- *Pessimism and negative thinking patterns
- *Anger, aggression, restlessness, irritability, tantrums, defiance
- *Constant worry about things that might happen or have happened
- *Crying
- *Physical complaints such as stomachaches, headaches, fatigue
- *Avoidance behaviors
- *Sleeping difficulties
- *Perfectionism
- *Excessive clinginess and separation anxiety
- *Procrastination
- *Poor memory and concentration
- *Withdrawal from activities and family interactions
- *Eating disturbances

<http://www.kathyeugster.com/articles/article004.htm>

Self-Regulation

The primarily voluntary regulation of cognition, behavior, emotion and physiological states for the purpose of goal-directed actions

But it needs to be developed and learned

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Self-regulation of pain

- Pain anxiety and pain catastrophizing
- Tension, distress, nervousness, irritability
- The role of stress
- Changing one's thoughts and expectations
- Meditation
- Distraction

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This is self-regulation of sensory systems



HOW?

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Help with self-regulation

- Making life more regulated
 - Routine
 - Predictability



Thanks to my Lab

- Megan Schmittel
- Bree Kaufman
- Shanti Madhavan-Brown
- Shelby Muhn
- Anna Weatherly
- Gretchen Imel
- Rebecca Jokinen
- Debora Ferreira
- Rachel Wilson
- Blair Tiseo
- Aaron Hartshorne

www.chsbs.cmich.edu/CHARGE

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Professional Day Poster

The CHARGE Syndrome Research Lab at Central Michigan University

Timothy S. Hartshorne, Ph.D.

Presenter Information

Tim Hartshorne is a professor of psychology, specialized in school psychology, at Central Michigan University. He is the grant holder for DeafBlind Central: Michigan's Training and Resource Project, which provides support to children who are deafblind in Michigan. Much of his work is influenced and motivated by his son Jacob, who was born in 1989 with CHARGE syndrome. Tim's particular interests include understanding the challenging behavior exhibited by many individuals with deafblindness, CHARGE, and related syndromes, and also how severe disability impacts the family. He is the lead developer of a deafblind intervener training module on behavior for the National Center on Deaf-Blindness. He has been awarded the Star in CHARGE by the CHARGE Syndrome Foundation. His research was recognized in 2009 with the Central Michigan University President's Award for Outstanding Research. He is a frequent presenter on CHARGE and deafblindness.

Co Presenters: Megan Schmitt, Bree Kaufman, Shanti Madhavan-Brown, Anna Weatherly, Shelby Muhn, Rachel Wilson, Gretchen Imel, Blair Tieso, Aaron Hartshorne


Presentation Abstract

The CHARGE Lab at Central Michigan University began about 1999 when Tim Hartshorne began to study behavior in children with CHARGE. This presentation provides a bit of history, and then describes the research of the current lab members.



Trying to understand the behavior


- It's because they are deafblind
- It's because they have communication problems
- It's because of vestibular difficulties
- It's because of early experiences
- Early research on
 - Executive function
 - Autistic like behaviors
 - Attachment
 - Sleep disturbances
 - Psychiatric diagnoses and medication



CHARGE
Research Lab

Current Projects

- Development of Play
- Recreation
- Medicine side effects
- Fun Chi
- Profiles of ability in CHARGE
- Anxiety and calendar systems
- Restorative Environments
- Headaches




Scottsdale CHARGE conference

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Current Lab

Maria Ramirez
Ben Kennert
Megan Schmittell
Bree Kaufman
Shanti Brown
Shelby Muhn
Anna Weatherly
Lily Slavin
Gretchen Imel
Becca Jokinen
Debora Ferriera
Rachel Wilson
Blair Tiseo
Aaron Hartshorne



<https://www.cmich.edu/colleges/chsbs/Psychology/charge>
Facebook: CHARGE syndrome research lab



Professional Day Poster

Milestones in CHARGE Syndrome

Meg Hefner, MS, Saint Louis University
Emily Fassi, MS, Washington University in St. Louis
Kevin Ballard

Presenter Information

Meg is a genetic counselor with 35 years of experience with CHARGE syndrome. She was a founding Board member of the CHARGE Syndrome Foundation and remains active as an Advisor to the Board. She has written extensively on CHARGE for families (Management Manual for Parents), geneticists (American Journal of Medical Genetics special issue on CHARGE syndrome), and professionals working with children with CHARGE (CHARGE Syndrome, Plural Publishing). Awards for CHARGE syndrome work include the first Stars in CHARGE award and the Diane Baker Alumni Award from the University of Michigan. She has presented at every CHARGE Syndrome Conference and actively answers genetic and medical questions by email and on several CHARGE Facebook pages. Meg's most recent endeavor is the CHARGE syndrome database (CSCDP), created to facilitate research by providing baseline information on individuals with CHARGE to researchers around the world.

Presentation Abstract

CHARGE Syndrome Clinical Database Project is a web-based, parent-completed database and registry with more than 1000 data items of medical and developmental features. Participants may upload photographs and limited medical records. Data are exported to Excel for analysis. More than 150 participants have entered relatively complete data. Although some participants are young and therefore have not yet reached milestones, we have data on around 100 for most milestones. Multiple milestones will be presented.



Professional Day Poster

CHARGE syndrome: the importance of inner ear MRI and CHD7 testing for diagnosis as the dysmorphology is not always obvious

Meg Hefner, MS

Presenter Information

Meg is a genetic counselor with 35 years of experience with CHARGE syndrome. She was a founding Board member of the CHARGE Syndrome Foundation and remains active as an Advisor to the Board. She has written extensively on CHARGE for families (Management Manual for Parents), geneticists (American Journal of Medical Genetics special issue on CHARGE syndrome), and professionals working with children with CHARGE (CHARGE Syndrome, Plural Publishing). Awards for CHARGE syndrome work include the first Stars in CHARGE award and the Diane Baker Alumni Award from the University of Michigan. She has presented at every CHARGE Syndrome Conference and actively answers genetic and medical questions by email and on several CHARGE Facebook pages. Meg's most recent endeavor is the CHARGE syndrome database (CSCDP), created to facilitate research by providing baseline information on individuals with CHARGE to researchers around the world.

Presentation Abstract

CHARGE syndrome (CS) is a complex and extremely variable genetic condition, most often caused by mutations in the CHD7 gene. The diagnosis still remains primarily clinical. Baby A is a subtle case where the diagnosis would likely have been greatly delayed without MRI and CHD7 testing. At birth, she did not have unusual ears or any of the other dysmorphic features typical of CHARGE.



Professional Day Poster

A new feeding scale for use in CHARGE syndrome

Alexandra Hudson, MD Candidate 2018
Dr. Kasee Stratton and Dr. Kim Blake

Presenter Information

Alexandra is a medical student at Dalhousie University in her third year of study. Prior to attending medical school, she completed my Honors Bachelor of Science at the University of Toronto in human biology, physiology, and psychology. She is currently studying CHARGE syndrome and its associated eating and gastrointestinal issues, under the supervision of Dr. Kim Blake at the I.W.K. Hospital in Halifax, Nova Scotia.

Presentation Abstract

Feeding issues are a major cause of morbidity and mortality in CHARGE syndrome. The feeding difficulties are unique and are not accurately captured with current validated feeding scales. This study designed a new feeding scale specific for CHARGE syndrome, and tested its validity and reliability in 100 individuals. This new scale can be used to assess an individual's current severity of feeding difficulties, track their oral feeding progress, and can alert to areas of concern that need clinical intervention.

Feeding Assessment for CHARGE Syndrome (FACS) Scale

Date: _____

Name of Individual with CHARGE Syndrome: _____

Age: _____

Gender (Circle one): Male

Female

Not Disclosed

Completed By (Circle one): Mother

Father

Feeding Therapist

Nurse/Physician

Other: _____

What percentage of your child/adult's daily fluid/nutrition intake is by G/J tube feeding? (Circle one percentage):

0%	25%	50%	75%	95%
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Circle one number on the scale:		Never	A Little	Sometimes	A lot	Always
1	He/she will refuse food when eating orally.	0	1	2	3	4
2	He/she takes longer than 45 minutes to eat orally.	0	1	2	3	4
3	He/she takes less than 15 minutes to eat orally.	0	1	2	3	4
4	He/she needs close supervision when eating orally.	0	1	2	3	4
5	He/she needs someone in the room when eating orally.	0	1	2	3	4
6	He/she has problems cutting food when eating orally.	0	1	2	3	4
7	He/she has problems feeding him/herself when eating orally.	0	1	2	3	4
8	He/she chokes or coughs when eating orally.	0	1	2	3	4
9	He/she has trouble chewing food.	0	1	2	3	4
10	He/she has trouble swallowing food.	0	1	2	3	4
11	He/she has to be told or reminded to chew.	0	1	2	3	4
12	He/she has to be told or reminded to swallow.	0	1	2	3	4
13	He/she does not like to mix food textures when eating (e.g. mixing puree and solid food).	0	1	2	3	4
14	He/she accidentally loses food out of his/her mouth during eating.	0	1	2	3	4
15	He/she will over-stuff his/her mouth with food during eating.	0	1	2	3	4

Circle one number on the scale:		Never	A Little	Sometimes	A lot	Always
16	He/she has difficulty moving food around with his/her tongue during eating.	0	1	2	3	4
17	He/she has a hard time feeling food or anything touching the inside of his/her mouth.	0	1	2	3	4
18	He/she dislikes oral eating.	0	1	2	3	4
19	He/she lets food sit in his/her cheeks or palate during eating (on purpose or not).	0	1	2	3	4
20	He/she will have food hidden in his/her cheeks or palate after the meal has ended (on purpose or not).	0	1	2	3	4
21	The Parent/Caregiver gets worried about their child/adult's ability to eat orally.	0	1	2	3	4
22	The Parent/Caregiver has difficulties feeding their child/adult. (e.g. preparing food the right way, getting enough information about helping them eat/drink)	0	1	2	3	4
Does the child/adult have problems with:		No		Yes		
23	Cold foods	0		1		
24	Room temperature foods	0		1		
25	Warm foods	0		1		
26	Thin liquids (e.g. water)	0		1		
27	Pureed foods (e.g. applesauce)	0		2		
28	Mashed lumpy food (e.g. mashed potatoes or mashed vegetables)	0		2		
29	Soft chewable foods (e.g. bread, crackers)	0		2		
30	Tough chewable foods (e.g. meat)	0		1		
31	Hard vegetables and fruit (e.g. raw apples)	0		1		

Total Score (sum of all items)	/100 total points
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Questions (1-4)

1. Which description best describes your child/adult's **current** feeding difficulties? (Circle one)

0 – **Very mild.** (E.g. Feeding difficulties are not an issue, doesn't need any feeding therapy, can handle all foods, can eat while alone, no trouble with chewing or swallowing)

1 – **Mild.** (E.g. Can handle most food, may have some feeding difficulties once in a while)

2 – **Mild to moderate.** (E.g. Can handle most food, may have some chewing or swallowing difficulties, may need some feeding therapy)

3 – **Moderate.** (E.g. Needs supervision sometimes when eating, has trouble chewing or swallowing certain foods, has feeding difficulties most of the time)

4 – **Severe.** (E.g. needs feeding therapy, needs assistance when eating, needs supervision when eating, has trouble with many food textures,)

5- **Very severe.** (E.g. Has a lot of feeding difficulties, needs feeding therapy, has trouble chewing and swallowing, has trouble with lots of food textures, chokes when eating, pockets food in cheeks, overstuffing mouth when eating)

2. How well do you think your child's score on this questionnaire describes the severity of their **current** feeding difficulties?

0 – Not at all.

1 – Poorly.

2 – Only a little bit.

3 – Well

4 – Very well

5 – Extremely well

3. Do you have any suggestions for improvement of these questions in this questionnaire? (Please describe)

4. Do you think this questionnaire has missed any important questions about feeding problems in CHARGE syndrome? (Please describe)



Professional Day Poster

Overview of Sexuality Education

Chathuri Illapperuma, Jordan Parker, Emily Mathis, Mallory Eddy, Alex Clarke, Kasee Stratton

Presenter Information

Chathuri Illapperuma, Jordan Parker, Emily Mathis, Mallory Eddy, and Alex Clarke are graduate students in the school psychology program at Mississippi State University and have been active members in the Bulldog CHARGE Syndrome Research Lab led by Dr. Kasee Stratton since September 2015. As a group, we research sexuality education for CHARGE in order assist and educate those individuals closely associated with the syndrome.

Presentation Abstract

The quality of sexuality education for students receiving special education has been an area of concern. Absence of federal regulations and a universal, accessible curriculum and learning outcomes may leave some individuals with inadequate knowledge. Limiting access to sexuality education increases risk for vulnerable, unhealthy sexual activities, and lack of knowledge on safe and healthy relationships including intimate relationships. This poster aims to highlight the trend in sexuality education and the need for a consistent and holistic approach.



Professional Day Poster

Trends in and Barriers to Recreation Participation in CHARGE Syndrome

Gretchen Imel and Timothy S. Hartshorne, Ph.D.

Presenter Information

Gretchen Imel is a senior with a dual major in therapeutic recreation and psychology at Central Michigan University. She has been a member of the CHARGE Syndrome Research Lab for three years.

Presentation Abstract

Participation in recreational activities is a component of an optimal quality of life. However, individuals with CHARGE face many barriers to recreation. This study found that individuals with CHARGE are actively participating in a wide range of recreational activities with walking, swimming, and listening to music having the most participation and swimming, listening to music, and walking being rated as the top favorites. Barriers were identified as expense of activity and adaptive recreation equipment.



Professional Day Poster

Profiles of Ability in CHARGE Syndrome

Bree Kaufman, M.A. and Timothy S. Hartshorne, Ph.D.

Presenter Information

Bree Kaufman is a 4th year doctoral student in school psychology at Central Michigan University. She works with Dr. Tim Hartshorne in the CHARGE Syndrome Research Lab. Her current research focuses on degrees of functioning within CHARGE syndrome.

Presentation Abstract

Parents often wonder if their child's CHARGE is mild, moderate, or severe. The ABILITIES Index provides a way to profile the functional ability of persons with disability in 9 major domains: audition, behavior and social skills, intellectual functioning, limbs, intentional communication, tonicity, integrity of physical health, eyes, and structural status. The purpose of this research was to explore the usefulness of the ABILITIES Index for CHARGE.



Professional Day Poster

Modelling hypoventilation and anesthetic recovery in a zebrafish (*Danio rerio*) model of CHARGE syndrome

Jessica MacLean

Presenter Information

Jessica is a first year medical student at Dalhousie University and is working on a project investigating adverse events following anesthesia administration in individuals with CHARGE syndrome using a zebrafish animal model. Her research is supervised by Dr Kim Blake and Dr Jason Berman and is conducted in the Berman lab which has a zebrafish model of CHARGE syndrome. Jessica completed a Master's in Science during which she conducted research in the area of cardiovascular pharmacology before entering medical school. She is interested in both the clinical and research aspects of CHARGE syndrome, and is looking forward to being able to take what is seen clinically and model it in the lab in hopes of furthering the understanding of the mechanisms of CHARGE.

Presentation Abstract

CHARGE patients commonly experience both minor and severe adverse events following anesthesia and sedation administration. There can be increased mortality and morbidity resulting from abnormal heart rate and decreased respiratory rate. Zebrafish that have been genetically engineered to lack *chd7* exhibit features of CHARGE. We will examine responses to anesthetic agents in this zebrafish model in an attempt to investigate causative factors. We will also investigate the regulation of *phox2b*, a gene involved in controlling respiration.



Professional Day Poster

Moving forward with the transition to preschool through collaborative planning

Julie Maier, Educational Specialist
Myrna Medina, Family Engagement
Specialist for California Deafblind
Services

Presenter Information

Julie Maier is an Educational Specialist for California Deafblind Services and provides technical assistance to families of children with deafblindness and their school teams. She has served many children and teens with CHARGE syndrome. Julie also serves as the Program Coordinator for an OSEP funded personnel preparation project "Specialization Program in the Education of Learners who are Deafblind " at San Francisco State University. Julie has been involved in the field of special education in the area of moderate-severe disabilities since 1987, including as a faculty member of SFSU Special Education Department since 1999. Julie has presented at numerous conferences on the topics of meaningful assessment for children with deafblindness and training providers in effective educational practices, including 2015 International TASH Conference, 2015 International CHARGE Syndrome Conference, and 2016 German CHARGE Syndrome Conference. She has written or co-authored several published articles on assessment, self-determination, social supports, collaborative partnerships between families and school teams during the transition process.

Myrna Medina has served as CDBS Family Engagement Specialist since 2000 supporting families of children with deafblindness. She is the parent of two children, one of whom is deafblind as a result of an unknown etiology. Myrna is a native of Mexico and fluent in English and Spanish. She has been instrumental in ensuring linguistic and cultural sensitivity of all CDBS Spanish language services and products. Myrna serves as the coordinator and as an active regional participant of the CDBS Network of Family Support Providers and is currently serving her second term as Secretary of the Coalition of Parents and Educators Deaf-Blind, a statewide family organization on behalf of children who are deafblind. Myrna also plays a leadership role in numerous organizations throughout the state, including Unidad y Fuerza (the FRC at Miller Children's Hospital in Long Beach), Centro de Niños y Padres at California State University Los Angeles, Birth-to-Five Vision Network, Junior Blind of America, and Dicapta, an OSEP-funded accessible communication development project. Myrna presents on numerous topics including: improving team collaboration and partnerships; sibling issues; working with Spanish speaking families; and communication between home and school. She presented at the Cal-TASH 2005, CEITAN, Fiesta Educativa and International CHARGE Syndrome Conferences 2013 and 2015. She has written and co-authored fact sheets and published articles on the importance of maintaining collaborative relationships between home and school, transition, and team work. She is also the coordinator for the national monthly CHARGE calls, and she acts as the co-presenter for the quarterly Spanish Webinars sponsored by NCDB, NFADB and CDBS.

Presentation Abstract

Times of transition can be one of the most anxious and stressful periods in a family's life, especially that first transition from early intervention services to preschool. We will share suggestions, tips, and resources families can use to be actively involved in the transition assessment process and strategies which early interventionists, educators and service providers can employ to encourage and support the family's involvement and ensure a smooth and successful transition to preschool services.



Professional Day Poster

Decision-making in the removal of gastrostomy tubes or buttons in CHARGE syndrome

Rachel Malta and Timothy S. Hartshorne, Ph.D.

Presenter Information

Rachel Malta recently completed her specialist degree at Central Michigan University, and is employed as a school psychologist in Charlotte, Michigan. While at CMU she was a member of the CHARGE Syndrome Research Lab.

Presentation Abstract

Gastrostomy tubes are often used to maintain adequate nutrition, but within the CHARGE population many parents eventually decide to have the tube removed. A survey was distributed addressing parental support needs and decision reasoning regarding removal. Results suggested a trusting, collaborative relationship between medical professionals and parents of the child, and clear, published guidelines regarding the process and timeline would be useful. Parents reported consulting a variety of professionals and parents with similar situations.



Professional Day Poster

Let's Talk About Sex: Implications for Future Research in Sexuality Education

Emily Mathis, Alex Clarke, Mallory Eddy, Chathuri Illapperuma, Kasee Stratton

Presenter Information

Emily Mathis, Alex Clarke, Mallory Eddy, Chathuri Illapperuma are second year School Psychology graduate students at Mississippi State University and members of the Bulldog CHARGE Syndrome Research Lab. Collectively, they work on the Sexuality Research Team. Sexuality education is crucial for teaching safety and relationship knowledge and this team was formed in response to the lack of sexuality education for individuals who have CHARGE or are deaf-blind. They hope to bring awareness and education to families and individuals with CHARGE, while working to develop a comprehensive sexuality education appropriate for individuals who are deaf-blind.

Presentation Abstract

Current literature on Comprehensive Sexuality Education (CSE) programs for individuals who are deafblind indicates CSE is beneficial. Such programs increase awareness of sexually transmitted diseases, promote safety, and improve general knowledge of development. However, there is limited research and resources available for individuals who have multisensory impairments. This poster aims to provide areas for future research by addressing deficits in the literature. A study proposal to expand sexuality education for CHARGE will also be presented.



Professional Day Poster

Post-Traumatic Growth in Parents of Children with CHARGE

Shelby Muhn and Timothy S. Hartshorne, Ph.D.

Presenter Information

Shelby Muhn is a first-year school psychology doctoral student at Central Michigan University. She has been a member of the CHARGE Syndrome Research Lab at Central Michigan University since April of 2016.

Presentation Abstract

Having a child with a severe medical and physical disability can be a traumatic event for families to face. They may feel isolated, guilty, and have moments of discouragement as they experience challenges. Although they face these encounters many parents begin to demonstrate considerable strength. The current research is looking to see what contributes to the phenomenon of post-traumatic growth in parents of children with CHARGE. Future goals include interventions and therapies to facilitate growth in parents.



Professional Day Poster

Examining the Measurability and Attainability of IEP Goals for Individuals with CHARGE Syndrome

Hailey E. Ripple, Johnna Dowdy, Olivia Kesler,
Katherine Johnson, Kasee Stratton

Presenter Information

Hailey is a doctoral student in the school psychology program at Mississippi State University and has been an active member in the Bulldog CHARGE Syndrome Research Lab led by Dr. Kasee Stratton. Hailey's research interests related to CHARGE include researching IEPs and parent perceptions of the IEP process, behavior, and strategies to raise awareness for CHARGE. Hailey was also one of the first Davenport Fellows.

Presentation Abstract

In order for an individual with a special education ruling to make appropriate educational and social progress in school, it is critical that the goals included in their individualized education programs (IEP) are measurable and attainable. The current study aims to examine the previously mentioned variables in IEPs written for individuals with CHARGE Syndrome. IEPs for individuals of various ages are included in results. Best practice in writing IEP goals will be included.



Professional Day Poster

IEP Transition Plans for Individuals with CHARGE Syndrome

Hailey E. Ripple, Elizabeth Struna, Matthew Ferrigno, Katherine Johnson, Kasee Stratton

Presenter Information

Hailey is a doctoral student in the school psychology program at Mississippi State University and has been an active member in the Bulldog CHARGE Syndrome Research Lab led by Dr. Kasee Stratton. Hailey's research interests related to CHARGE include researching IEPs and parent perceptions of the IEP process, behavior, and strategies to raise awareness for CHARGE. Hailey was also one of the first Davenport Fellows.

Presentation Abstract

Transition plans included on individual education programs (IEPs) can be an unfamiliar topic for families when it comes time to plan for an individual's future after school. Transition plans aim to guide individuals towards opportunities for further education, employment, and independent living (Trainor, Morningstar, & Murrar, 2016). Current data on transition plans for individuals with CHARGE syndrome will be presented. Researchers aim to educate parents about necessary components of transition plans and ways to ensure that post-school goals are achieved.



Professional Day Poster

Social Play in Children with CHARGE Syndrome

Megan C. Schmittell and Timothy S. Hartshorne, Ph.D.

Presenter Information

Megan Schmittell is a fourth year graduate student in the school psychology doctoral program at Central Michigan University. She is a member of The CHARGE Syndrome Research Lab at Central Michigan University, and her research is focused on social play in children with CHARGE syndrome.

Presentation Abstract

Play is a fundamental aspect of development. As children age, they progress through different stages of social play supporting their social, emotional, and cognitive development. There is little information on how children with CHARGE syndrome develop social play. The current poster reports results from a study documenting the stages of social play in which a sample of children with CHARGE syndrome are functioning, as well as, information regarding their play preferences, as reported by parents.



Professional Day Poster

Research on Headaches in Children with CHARGE Syndrome

Lily Slavin and Timothy S. Hartshorne, Ph.D.

Presenter Information

Lily Slavin is a recent graduate of Alma College with a psychology major. Through a summer research program and independent study she worked in the CHARGE Syndrome Lab at Central Michigan University during the summer and fall of 2016.

Presentation Abstract

This poster will summarize the results of our study on the presence and incidence of headaches in CHARGE syndrome. We looked for associations between the presence or absence of CHARGE characteristics and headaches. We also looked at the presence of headaches and sleep, hormone replacement therapy, and eating. Finally, we investigated the behaviors used by parents to identify headaches.



Professional Day Poster

The Bulldog CHARGE Syndrome Research Lab at Mississippi State University

Kasee Stratton, Ph.D., NCSP

Presenter Information

Dr. Kasee Stratton is an assistant professor of school psychology at Mississippi State University. She is also a licensed psychologist and nationally certified school psychologist. She directs the Bulldog CHARGE Syndrome Research Lab at MSU. Dr. Stratton has been researching and presenting about CHARGE since 2005. She is an author of two chapters in the book, CHARGE Syndrome, the developer of the CHARGE Non-Vocal Pain Assessment, and has presented in the U.S., Australia, New Zealand, and Denmark on CHARGE. Dr. Stratton specializes in challenging behavior and improving adaptive skills.

Presentation Abstract

The Bulldog CHARGE Syndrome Research Lab at Mississippi State University conducts research and helps to spread awareness of CHARGE. The lab is composed of undergraduate students and graduate students in school psychology. The lab is under the direction of Dr. Kasee Stratton. Our goal is to investigate areas that are important to families, professionals, and individuals with CHARGE. Additionally, the lab seeks to create a diverse group of advocates and future professionals who are well informed of CHARGE syndrome.



Professional Day Poster

The CHARGE IEP: What Accommodations and Rulings are Common?

Elizabeth Struna, Hailey Ripple, Amanda Cosgriff, Whitney Driskell, Savannah Trice, Tierra Kilbert and Kasee Stratton

Presenter Information

Elizabeth Struna is a first year Doctoral student in the School Psychology program at Mississippi State University. Elizabeth has been a member of the Bulldog CHARGE Research Lab since the Fall Semester of 2016. Since joining the Lab, she has aided with the research of Individualized Education Plans (IEPs) in CHARGE. Elizabeth has always shown an interest in school compliance with IEPs. By assisting with this study, Elizabeth hopes to bring together educators and parents.

Presentation Abstract

Although CHARGE syndrome has a diverse presentation, little is known about the common educational accommodations or services provided for students with CHARGE. Yet, many children with CHARGE share a common special education category—deafblind. This poster will review the most frequent special education accommodations and services, as well as highlight the special education categories by which students are served. This information should assist professionals and parents in advocating for students with CHARGE and will also serve to identify any underserved educational areas.



Professional Day Poster

What makes up a family in CHARGE: Family Dynamics and Demographics

**Madison Sully, Megan Anderson,
Kasee Stratton,**

Presenter Information

Madison Sully is an educational specialist graduate student in the School Psychology program at Mississippi State University (MSU). Madison is interested in family dynamics and relationships in CHARGE families. Madison joined the Bulldog CHARGE Syndrome Research lab at MSU upon her first year of graduate school in 2014. CHARGE quickly became an interest of Madison's due to the limited information available. As a researcher on the family dynamics and relationships team, Madison is interested in understanding the dynamics of families who have a child with CHARGE Syndrome. In 2015, Madison attended her first CHARGE Conference in Chicago. The experience of meeting many individuals with CHARGE and their families has brought her back to her second conference with more research to share.

Presentation Abstract

Our research is intended to understand the make up and dynamics of families that include a child with CHARGE Syndrome. At present, no research has reviewed the family demographics or parental relationships in CHARGE Syndrome. The researchers seek to understand the differences and similarities between families with a child with CHARGE. Additional research questions addressed will include the number of children with CHARGE, birth order, and divorce rates.



Professional Day Poster

Adverse Effects and Behavioral Analysis of Psychotropic Drug Use with Children with CHARGE Syndrome

Blair Tiseo and Timothy S. Hartshorne, Ph.D.

Presenter Information

Blair Tiseo is a senior studying neuroscience at Central Michigan University. She has been in the CHARGE research lab for one year and focuses on psychotropic medications in those with CHARGE syndrome. Blair is planning to go to graduate school in spring of 2018 to become a Physician's Assistant.

Presentation Abstract

The use of psychotropic medications in the CHARGE population is understudied in relation to adverse effects, outcomes, and usage. A single-subject design will be used to study individuals with CHARGE syndrome who are beginning a psychotropic medication prescribed by their physician. The results will show the relationship between adverse effects and symptoms before starting the regiment. This study will show the adverse effects specific to those with CHARGE syndrome and whether or not the medications are effective.



Professional Day Poster

Restorative Environments

Anna Weatherly and Timothy S. Hartshorne, Ph.D.

Presenter Information

Anna Weatherly is a first-year school psychology doctoral student at Central Michigan University. She has been a member of the CHARGE Syndrome Research Lab at Central Michigan University since August of 2016. She also holds a Master's Degree in Environmental Psychology from the University of Surrey.

Presentation Abstract

Mental fatigue, or the reduced ability to block distractors and attend to important stimuli, has many cognitive and behavioral consequences, such as, difficulties in problem solving, a tendency to act rashly, being irritable, less patient, uncooperative, and problems making and following plans. According to Attention Restoration Theory, spending time within a restorative environment is the best way to recovery from mental fatigue. Beginning to understand how individuals with CHARGE Syndrome experience mental fatigue and restorative environments may be beneficial.



Professional Day Poster

Issues in Psychotropic Medication Use for Children with CHARGE Syndrome

Rachel Wilson and Timothy S. Hartshorne, Ph.D.

Presenter Information

Rachel is a junior at Central Michigan University. Currently, she is working on finishing a degree in psychology and is involved in the CHARGE lab on campus led by Tim Hartshorne. Her project is focused on psychotropic medications used on children with CHARGE syndrome.

Presentation Abstract

Psychotropic medications are often prescribed to children with CHARGE as a way to reduce challenging behaviors. Issues concerning prescribing children with CHARGE psychotropic medications include lack of prospective research done on long term effects of these medications, off-label prescribing, side effects, and the effect these drugs have on the child's ability to communicate.