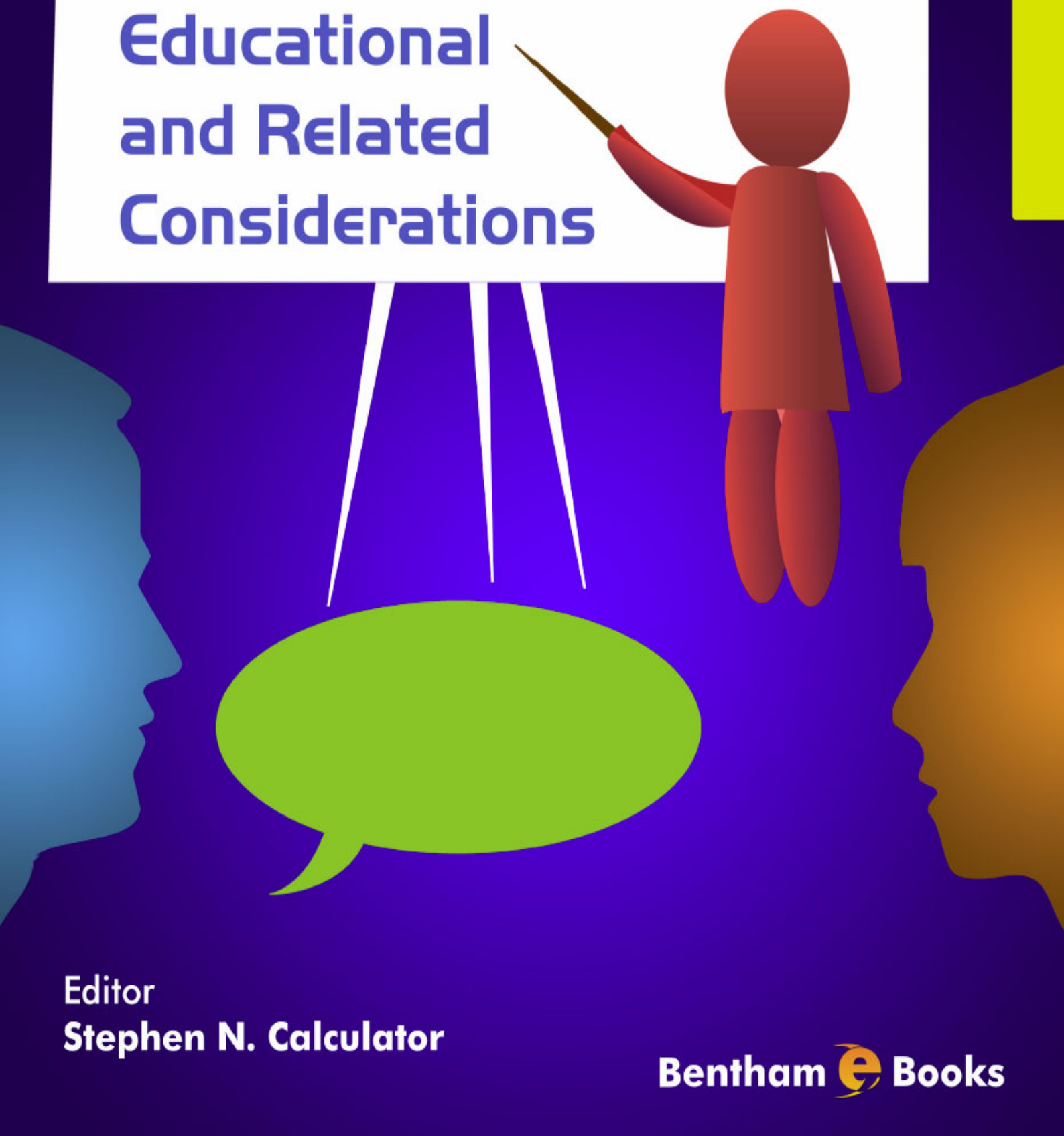
eISBN: 978-1-68108-116-8
ISBN: 978-1-68108-117-5

Angelman Syndrome: Communication, Educational and Related Considerations



Editor
Stephen N. Calculator

Bentham e Books

ANGELMAN SYNDROME: COMMUNICATION, EDUCATIONAL AND RELATED CONSIDERATIONS

Edited By:

Stephen N. Calculator
Department of Communication Sciences & Disorders
University of New Hampshire (USA)

advertisements or ideas contained in the Work.

Limitation of Liability:

In no event will Bentham Science Publishers, its staff, editors and/or authors, be liable for any damages, including, without limitation, special, incidental and/or consequential damages and/or damages for lost data and/or profits arising out of (whether directly or indirectly) the use or inability to use the Work. The entire liability of Bentham Science Publishers shall be limited to the amount actually paid by you for the Work.

General:

1. Any dispute or claim arising out of or in connection with this License Agreement or the Work (including non-contractual disputes or claims) will be governed by and construed in accordance with the laws of the U.A.E. as applied in the Emirate of Dubai. Each party agrees that the courts of the Emirate of Dubai shall have exclusive jurisdiction to settle any dispute or claim arising out of or in connection with this License Agreement or the Work (including non-contractual disputes or claims).
2. Your rights under this License Agreement will automatically terminate without notice and without the need for a court order if at any point you breach any terms of this License Agreement. In no event will any delay or failure by Bentham Science Publishers in enforcing your compliance with this License Agreement constitute a waiver of any of its rights.
3. You acknowledge that you have read this License Agreement, and agree to be bound by its terms and conditions. To the extent that any other terms and conditions presented on any website of Bentham Science Publishers conflict with, or are inconsistent with, the terms and conditions set out in this License Agreement, you acknowledge that the terms and conditions set out in this License Agreement shall prevail.

Bentham Science Publishers Ltd.
Executive Suite Y - 2
PO Box 7917, Saif Zone
Sharjah, U.A.E.
Email: subscriptions@benthamscience.org

CONTENTS

FOREWORD

I was both pleased and honored to be asked to write the Foreward for this book – pleased because of the importance of the book's topic, and honored because of my admiration for Steve Calculator's many contributions to the field of augmentative and alternative communication (AAC). Steve and I first met in 1993 when we were invited to present a paper at the same conference he describes in the book's Preface. The paper was focused on best practices for the delivery of AAC services in inclusive educational settings and was part of the second National Symposium on Effective Communication for Children and Youth with Severe Disabilities in Washington DC. Actually, the truth of the matter was that Steve was invited, and he invited me. I was flattered and more than a little intimidated by his invitation, because he had already established himself as a leader in the AAC area and I was a relative newcomer to the field. However, any trepidation I experienced was eradicated once we began our long distance collaboration, and as I came to know Steve as one of the most open-minded and open-hearted colleagues with whom I have ever worked. Those qualities are reflected in the team of authors he brought together in this book, representing parents, clinicians, researchers, and policy makers who live with, support, and advocate for individuals with Angelman syndrome (AS).

Why do we need this book? Anyone who has read the (very limited) literature on AS will come away believing that they "know" four things: (1) people with AS smile and laugh a lot, (2) very few are able to speak or acquire literacy skills, (3) most engage in frequent problem behaviors, and (4) we don't know much about what to do about (2) and (3). We need this book because these "facts" – these *inaccurate* facts -- lead to low expectations, and low expectations lead to poor educational and communication outcomes. In contrast, the authors in this book offer both solutions and hope as they describe practical, innovative solutions that are aimed at improving the lives of people with AS. We need this book because people with AS need us to know that they are not "happy puppets" – they are children, adolescents, and adults who are capable of learning and of making real contributions to their home and school communities. We owe Steve and his collaborators a debt of gratitude for compiling this collection of thoughtful, forward-looking chapters that will surely change the lives of people with AS and their families.

Pat Mirenda
Centre for Interdisciplinary Research and Collaboration in Autism (CIRCA)
University of British Columbia
Vancouver, BC V6T 1Z4 Canada

PREFACE

Approximately 25 years ago, I found myself speaking at a conference in Washington, D.C. that was intended to delineate priorities for federal funding of research related to the education of students with severe disabilities. As I left the stage and made my way back to my seat, a member of the audience grasped my arm and pulled me towards her. She asked, "What do you know about Angelman Syndrome?" I indicated I had seen a few children with the syndrome, including twins, and had worked with their schools in developing communication, educational, and related programs. The woman beamed and proclaimed, "Then you are an expert in Angelman Syndrome." I reiterated my limited contact with these individuals but undeterred, she repeated, "You're an expert – we want you to help our school develop a program for our daughter in Virginia."

I accepted the invitation and agreed to travel to Virginia to assist the school team in any way they might find helpful. Prior to the visit, I searched EVERYWHERE for information about Angelman Syndrome. Other than descriptions of classic characteristics, and rather foreboding prognosis, there was little useful information. Ultimately, I found myself relying on my knowledge of the general population of individuals with severe disabilities, tweaking this understanding to accommodate special features associated with AS.

I surmised the visit to Virginia went well when I was soon contacted by another parent, then another, and another. Professionals reached out as well, all looking for possible answers to conundrums presented by these children and adults. As much as I wanted to help, I often found I had more questions than answers. My research focus eventually shifted to communication considerations for individuals with AS. Fortunately the University of New Hampshire encouraged its faculty to engage in outreach/consulting. Soon I was focusing exclusively on Angelman Syndrome.

Approximately three years into this journey, I was contacted to provide a consultation for a school-age boy in North Carolina. His parents corresponded with me prior to the visit to express their fear I would not see their child's potential and might instead reinforce his teachers' low academic expectations of him. The visit went quite well – we even articulated both a short and long-term vision for the student that accommodated a belief that with the right supports his potential was boundless.

This boy's Mom drove me to the airport and when I exited her car, she gave me an ornately wrapped present. She then departed. There was a major delay in my flight so, not being one who enjoys surprises, I opened the gift. It was a rather waxy, difficult to identify object in the general shape of an oblong. I scratched it, smelled it, and shook it but to no avail – I could not

identify it.

Upon arriving home in New Hampshire, I presented the object to my wife who conjectured it was soap. We wet it but nothing happened. Next, we hypothesized it was a candle but to no avail as there was no wick. We remained stumped.

When I returned to work a few days later, I had a message from the Mom on my voicemail, "Thank you for dreaming with us and not letting others short-change our son. We were once told our son was a VEGETABLE who could not learn and would end up in total custodial care. We hope you liked the "eggplant". Maybe it will serve as a reminder to others that no child is a vegetable." I keep that eggplant in clear sight in my office. It humbles me daily.

Individuals with Angelman Syndrome (AS) present a variety of challenges to educators, allied health professionals (*e.g.* speech-language pathologists, occupational therapists, and physical therapists), parents, and others. This book is intended to be a comprehensive resource that integrates theory with practical strategies aimed at optimizing individuals' acquisition of the types of functional skills that can contribute to positive changes in their overall quality of life. Given the complex nature of AS, it was determined that in order to properly meet this goal it was essential to address individuals' strengths and challenges from an international, multi-disciplinary perspective. As such, authors contributing to this book represent the USA, Canada, Australia, New Zealand, the Netherlands, Italy, Austria, and Sweden.

Readers will find that much of the content of this book has implications not only for individuals with AS but the broader population of individuals whose severe disabilities can be traced to other etiologies as well. However, the target group is indeed individuals with AS. With this in mind, authors were selected based on distinguished records of service to children and adults with AS. This includes not only substantive contributions to the scientific literature but also personal, hands-on clinical and educational experience.

The book begins with three prologues written by the parents of two children and one adult with AS. Parents were selected based on the editor's personal knowledge of their efforts in securing state-of-the-art educational, augmentative and alternative communication (AAC), and related services for their families as well as others. These parents were encouraged to use an informal writing style in sharing their perspectives in a manner that might enable other parents and professionals to glean helpful insights into the myriad of obstacles they faced and surmounted. These writers were encouraged to address 10 points, if possible: (a) their reactions to the diagnosis of AS; (b) initial dreams and nightmares for their children, and how these changed over time; (c) impact of AS on their families; (d) changes in the focus of their children's educational and other programs over time; (e) a description of their child's current life; (f) their perceptions of the future role they might play in ensuring their children the best

possible quality of life; (g) lessons learned along the way and corresponding advice they might like to share with other parents; (h) general take-away messages for professionals, including their expectations of these individuals; (i) their dreams for their children, and; (j) any concluding thoughts.

The book continues with chapter 1, Clinical Features, Medical Issues, and Diagnostic Testing in Angelman Syndrome, co-authored by Jennifer Mueller M.S., C.G.C., a Pediatric Genetic Counselor, and Charles Williams, M.D., Professor of Pediatrics, both from the Division of Genetics and Metabolism at the University of Florida. They share a special interest in neurogenetic syndromes and related conditions. This chapter reviews the clinical and medical aspects of AS, discussing genetic mechanisms that cause AS, reviewing clinical criteria, and exploring its natural history. Implications for learning and overall function are presented.

Chapter 2, Learning Characteristics of Students with Angelman Syndrome and Related Instructional Strategies, is written by Erin Sheldon, M. Ed., an Assistive Technology and Education Consultant specializing in emergent literacy, with a particular focus on students with AS, in Kingston, Ontario, Canada. She provides further discussion of clinical features of AS, emphasizing learning characteristics, their relevance in meeting students' educational needs, and implications for successful inclusion in academic instruction.

Stephen Calculator, Ph.D., Professor of Communication Sciences and Disorders at the University of New Hampshire, in Durham NH and member of the Scientific Advisory Committee of the Angelman Syndrome Foundation, is the author of Chapter 3, Broad Considerations Driving AAC and Related Instruction for Individuals With Angelman Syndrome. He discusses assessment and intervention considerations that are particularly well-suited to the learning characteristics of individuals with AS that were discussed in the previous chapters. Dr. Calculator cautions against over-reliance on standardized assessments, presenting several alternatives that flow continuously to intervention planning and implementation. The latter are discussed as a set of ecologically valid procedures that can be integrated into naturally arising, meaningful activities throughout the day.

Chapters 4, 5, and 6 focus on issues to consider when attempting to foster communication and related skills in individuals with AS. Chapter 4, Overview of Augmentative and Alternative Communication and Special Considerations for Individuals With Angelman Syndrome, written by Stephen Calculator, reviews pertinent concepts that are associated with the four primary components of AAC systems: (a) symbols, (b) techniques, (c) devices, and (d) strategies. AAC is discussed as a viable means of augmenting and/or when called for, replacing existing communicative behaviors. Both aided (*e.g.* communication devices) and unaided (*e.g.* signs and gestures) systems are considered.

In Chapter 5, Fostering Communication Skills in Beginning Communicators, the principal authors are Jeff Sigafoos, Ph.D., Professor of Education in the School of Educational Psychology and Pedagogy at Victoria University of Wellington in New Zealand, and Vanessa Green, Ph.D., Professor in the School of Education, also at Victoria University. Their distinguished group of co-authors are Robert Didden, Ph.D., Professor of Intellectual Disabilities, Learning, and Behavior at the Behavioral Science Institute and School of Education of the Radboud University Nijmegen, The Netherlands; Mark F. O'Reilly, Ph.D., Audrey Rogers Myers Centennial Professor in Education and Chair of the Department of Special Education at The University of Texas at Austin, TX, U.S.A.; Giulio E. Lancioni, Ph.D., Professor in the Department of Neuroscience and Sense Organs at the University of Bari in Bari, Italy; and Peter B. Marschik, Ph.D., Associate Professor of Physiology and Neurolinguistics at the Medical University of Graz in Graz, Austria and affiliated with the Karolinska Institute in Stockholm, Sweden. This chapter focuses on assessment and intervention considerations that relate directly to beginning communicators who rely primarily on pre-symbolic behaviors.

Next, in Chapter 6, Fostering Effective Communication Skills in Individuals With Angelman Syndrome Who Rely on Symbolic Methods of Communication, Stephen Calculator shifts the focus to individuals who are candidates for, and/or are already using, more advanced symbolic means of communication. Both unaided and aided methods are examined. Best practices corresponding to the four AAC components are delineated with an eye on promoting individuals' acceptance and successful use of AAC systems.

In Chapter 7, Enhanced Natural Gestures: A Self-Administered Program for Teaching Communicative Behaviors to Individuals With Angelman Syndrome, Stephen Calculator provides a tutorial on an unaided method of communication that has been observed to be particularly useful with individuals with AS, both beginning communicators and those using symbolic forms of communication. His tutorial is intended to enable teachers, parents, speech-language pathologists, and others to implement ENG instruction across multiple settings.

Chapters to this point include discussions of behavioral as well as more naturalistic approaches. Jane Summers, Ph. D., a psychologist and behavior expert at the Centre for Addiction and Mental Health in Toronto, Ontario, Canada, and member of the ASF Scientific Advisory Committee, authors Chapter 8, Application of Principles of Applied Behavior Analysis in Addressing Challenging Behaviors of Individuals With Angelman Syndrome, which ties these two approaches together. Among her many credits has been authoring and coordinating a series of practical, on-line modules for addressing challenging behaviors in individuals with AS, a project co-sponsored by the Canadian Angelman Syndrome Society (CASS) and the Angelman Syndrome Foundation (USA). Dr. Summers provides a highly

practical overview of how applied behavioral analysis can be used to replace problem behaviors with more appropriate methods of communication.

Chapter 9, Comprehensive Literacy Instruction for Students With Angelman Syndrome, transitions from communication to a specific focus on reading and writing. Erin Sheldon, who directs the Angelman Syndrome Literacy Project in Kingston, Ontario, Canada and Caroline Musselwhite, Ed.D., CCC/SLP, an Assistive Technology and Emergent Literacy Consultant in Phoenix, Arizona, who is a Fellow of the International Society for Augmentative and Alternative Communication (ISAAC), review principles of emergent literacy, highlighting specific adaptations, supports and opportunities that can be used to foster reading and writing skills in individuals with AS.

The book concludes with Chapter 10, The Role of AAC in Fostering Inclusion of Adults With Angelman Syndrome in Post-School, Home, and Community Settings. Co-authors Teresa Iacono, Ph.D., Professor of Rural Health and Regional Allied Health in the La Trobe Rural Health School at La Trobe University in Victoria, Australia and Hilary Johnson, Ph.D., Strategic Projects Advisor, Scope's Communication and Inclusion Resource Center, also in Victoria, discuss social interaction processes as a foundation for examining the use of AAC to enhance the inclusion of individuals with AS at home, work, and in the community.

If the primary goals of this book are met, readers should walk away with increased confidence in their abilities to foster communication, educational and related skills in individuals with AS across multiple environments over the life span. They should be able to identify as well as implement evidence-based practices supported by the most recent research. A posture of cautious optimism may be called for as top researchers across the world search for a cure for AS. Whether this amounts to a full reversal of disabilities associated with AS and/or reduced severity of some challenges but not others, remedial efforts will remain extremely important. Collectively, the authors of this book implore readers to take up this challenge with a hunger for new knowledge and the skills to implement it.

Stephen N. Calculator
Department of Communication Sciences and Disorders
University of New Hampshire, NH, USA

DEDICATION

This book was most definitely long in the making, reflecting educational and personal values spawned as a doctoral student under the mentorship of David Yoder in the University of Wisconsin's Department of Communicative Disorders back in 1980, when the "new" field of Augmentative and Alternative Communication was emerging. This set the stage for a professional career in research, teaching, and clinical/educational practice involving children and adults with severe disabilities. I owe a tremendous debt of gratitude to the hundreds of outstanding colleagues with whom I have collaborated, most recently and notably the distinguished group of authors who contributed to this book. Still, it has been parents and families in the Angelman community that have driven my passion to explore issues targeted in this book. They have reminded me on countless occasions that stakes and expectations for their children must be high. I also owe immeasurable gratitude to my own family – my wife Jean and four wonderful children, Bryant, Lauren, Trevor, and Kaitlin. They have provided a lifetime of reminders that the good life I have experienced must engender a commitment to do my part in ensuring the same for others.

PROLOGUE

A Collection of Three Essays

Plans, Paths and Possibilities

Eileen Braun[1]

Every person I meet on the path of this life's journey was placed here by God's divine purpose and plan. It is for me to seek out and understand, to learn and to determine how I will help them and how I will live my life given His purpose and plan. I may not know or find

all of the answers while I travel this earth, but I will continue the journey with determination, faith, compassion and charity, putting the needs of those who need help and guidance before my own wants, needs and desires. I will very likely not do this perfectly, some days I may fail, but I will always try. ~ Eileen Braun

My husband, Tom, and I have four beautiful children: Kaitlin-24, Joe-21, Jon-18 and Kim-16. Our oldest child, Kaitlin, has Angelman syndrome (deletion +). When Kaitlin was diagnosed with Angelman syndrome my reaction wasn't singular or one moment in time. It has been complicated and, I believe, one that has continued to evolve throughout her lifetime. I have loved Kaitlin since before she was born and I continue to love her more—and differently—every day.

To be honest, I knew right from her birth something was just not right. She didn't have that beautiful newborn baby cry. She yelled, but she didn't cry. She held her mouth open in a way other newborns didn't do and I could always see her tongue in the front of her mouth. There was no precious, sweet cooing. She had small, frequent foot tremors that happened more than they should. She had an exaggerated startle reflex. She was such a "messy" nurser, frequently losing out of her mouth as much breast milk as she'd swallowed. All of these signs on their own might be nothing, but together I knew something was not right.

Her pediatrician, whom I worked with in the NICU and PICU, very kindly and compassionately dismissed my concerns as that of a first-time mother and one who 'knew and had seen too much.' My gut told me differently, but I also wanted desperately to believe that our pediatrician was right. After all, we had done everything right. We had planned, we had prepared. Long before Kaitlin was conceived we were both physically healthy, had taken all the classes, read all the books, had a natural, drug-free delivery without complications, so there should be no reason that anything could go wrong.

Little by little, as people commented on what a good, quiet and content baby Kaitlin was, my gut remained uneasy. She rarely slept during the day and at night didn't sleep for more than an hour or two at a time. She was barely meeting her developmental milestones and by nine months looked like a baby diagnosed as failure-to-thrive. I was scared, confused and we weren't getting any support for our concerns—just watch and wait we were told.

Thinking we would lose this beautiful little girl, we pursued more aggressive assessments and testing. Ultimately this led us to a devastating [mis]diagnosis of Rett Syndrome when Kaitlin was 13 months old. I had never heard of this and I couldn't hear or comprehend at that instant what was being said. Nothing made sense. I was so angry and confused. How could something like this happen to this beautiful little girl—so sweet, and happy, and innocent? I was incredibly sad and devastated—not for Tom and me, but for Kaitlin; I could barely look

at her because my heart was broken for her. What would happen to Kaitlin—would she grow up, would she know us and have the capacity to love others and us? Would she walk, talk, and learn? Would others accept her or would they be cruel and tease her—I couldn't bear that for Kaitlin.

The following morning we met with the physician who had delivered the devastating diagnosis to us the day before. As much as I hated him the day before for dissolving our world, he told us that morning not what we wanted to hear, but what we needed to know: do not pity Kaitlin or feel sorry for her. That will not help Kaitlin. She is the same beautiful little girl she was before this diagnosis and he knew that we still loved her, so now we needed to roll up our sleeves, pull up our boot straps and get to work. Challenge her, make her work, have expectations that she can and will grow, develop and learn. At that point I had hope; I could take a breath and not crumble. We traveled back home and got right to work in therapies—physical, occupational, speech and aqua-therapy. Kaitlin was growing, progressing, gaining skills—she was learning and beginning to overcome some of her sensory issues. She was bright-eyed, engaging and full of life.

When Kaitlin was 18 months old, I attended a Rett Syndrome conference and as I cried and listened and learned I also again began to have hope that this was all wrong, that Kaitlin would be 'okay'. I discovered that Kaitlin was different than the other girls with Rett Syndrome. She wasn't regressing or losing skills—she continued to gain them! She didn't exhibit the typical symptoms of Rett. She was either atypical for Rett Syndrome or this was not the correct diagnosis. We were fortunate shortly after the Rett conference to be able to meet with experts in Rett syndrome to have them observe and assess Kaitlin. The conclusion: Kaitlin did not have Rett Syndrome—but they did not know what was causing Kaitlin's delays and they recommended we pursue further assessment and testing. Could this be? Should I allow myself to have hope that Kaitlin would be okay—that she would be "normal"?

We were referred to a neurologist at the University of Chicago and after meeting and assessing Kaitlin he agreed that she did not have Rett syndrome, but rather another very rare disorder—a disorder which he had only seen and diagnosed in one other patient—Angelman syndrome. He wrote the orders for the blood work for genetic testing, which would take six to eight weeks for the results, and then handed us a one-page document, not even a full page of information, about Angelman syndrome. He could tell us relatively little about this disorder. As Kaitlin was already in the therapies that would help her best, he had relatively little more to offer us. At that point I was stunned, hopeful, doubtful, numb. I didn't know what to think or what to hope for—would this be better or worse? If it was definitely Angelman, then we wouldn't get "normal" for Kaitlin. Admittedly, I was relieved Kaitlin did not have Rett syndrome, but I desperately did not want the diagnosis of Angelman syndrome—I wanted normal, typical, healthy little girl.

My mind and my heart went to that place where this could be fixed with therapy, diet and nutrition and perhaps a medication that would adjust some metabolic imbalance that was causing all of this—then Kaitlin would be okay and life would be back to normal. But that was not to be for Kaitlin, and at 21 months, the diagnosis of deletion of 15q.11-q.13 on the maternal 15th chromo-some—Angelman syndrome—was confirmed.

In 1991, the year Kaitlin was diagnosed, there was precious little information about Angelman syndrome. There was not the availability of information on websites, scientific or medical journal articles; no social media, chat groups or local or regional support groups—we were on our own. We ultimately found the Angelman Syndrome Foundation and attended our first conference in Orlando, Florida, in 1993. We were relieved to find and meet other families who were living—not simply surviving—life with Angelman syndrome. We were not alone.

From early on in our journey we embraced that people with Angelman syndrome have a great capacity to know and understand what is being said and happening around them. As Kaitlin's parents, if we believed this to be true for Kaitlin then we also maintained that if she had this capacity then she also had the capacity to love and be loved, to know and understand right from wrong, that she could and would learn, that she could and would be responsible for her actions and that she could and would continue to develop skills and acquire knowledge that, like her typically developing siblings, would help her to live as independent, productive and happy a life as possible. This is our hope and dream for all of our children, which most certainly includes Kaitlin! How this looks for Kaitlin has certainly evolved over these past 24 years, and will no doubt continue to change throughout her life.

The fact that Kaitlin has Angelman syndrome doesn't absolve her from accountability for her actions, nor does it give her special privileges or free passes on responsibility in the Braun family. We learned quickly that Kaitlin would allow us to do for her as much as we would do. When she was learning to walk we made her walk everywhere. It took longer—a lot longer—but we knew being tough with her would help her achieve the mobility and independence she needed. Over the years we've tried to modify very little in our home. Our perspective is that if we want Kaitlin to be able to travel with us, to go out to dinner and have good manners, to respect our and others' property and belongings, and to achieve success and independence outside of her and our home, then she had to be regularly exposed to and function in a typical routine and environment.

Raising a person with Angelman syndrome has certainly had profound effects on our family, both positive and challenging. To be honest, at almost every typical milestone and event that others observe and enjoy—school programs, sporting events, parties, sleep-overs, first date, first kiss, mother-daughter adventures, braces, driver's license, graduations, engagements,

weddings … the notion of 'what if' wants to creep in. What if Kaitlin didn't have Angelman syndrome? What would she/we be doing if…? How would she be in this situation if …? To this day it's not a place I venture often. It serves no purpose because our lives have been forever changed, not just by Kaitlin's diagnosis of Angelman syndrome, but with the addition of every child and every experience we as a family have had together. We can't undo what was or what has happened, but we can certainly build on and learn from our experiences.

Tom and I could not be more proud over the years to hear all of our children's teachers tell us how much they enjoyed having our children in their classroom; what good citizens they are in the classroom, caring, compassionate and considerate of others, respectful and their 'go-to' students when they or others need help. They have proven to be kind and caring young adults outside of school, hard working, responsible and accountable for their actions and decisions. I have no doubt they have learned this by observation, participation and necessity. They have been asked to do things that others who don't have a sibling with special needs would likely never understand, to help in situations they'd likely never volunteer for but do so to help their sister and parents out of kindness, love and devotion. They've been asked to give up time with one or both parents when we have had to 'divide and conquer' because the activity is such that Kaitlin can't or won't participate. Our children have also learned who their truest of friends are—those who know and respect Kaitlin, and our family, just as we are—and that has strengthened their bonds of friendship.

Kaitlin has helped us all to realize that what is truly important in life is that the focus is not on ourselves but in helping others. By shifting our priorities to focus and help others we have strengthened our commitment to our family. When we are focused on meeting the needs of others in the family then everyone's needs will be met.

Now that the bus has stopped coming for Kaitlin it is amazing to see how quickly those years flew by. As a parent I don't want to control or choose the life path for any of my kids, but with a daughter with Angelman syndrome, that is certainly what we have been tasked with doing for and with her. As we sat through countless team meetings and planning sessions, IEP meetings, parent-teacher conferences and many other meetings throughout Kaitlin's educational career, I always had in my mind—how will what we are doing here, today, get us to the day the bus stops coming? What environment is best suited for Kaitlin to learn, grow and thrive, not passively but actively participating in her educational day and learning experiences? How will this curriculum or activity benefit her now and is this a (n) experience, knowledge or skill that is necessary for her? Can we build on this toward life-long knowledge or skills? Will this help her achieve independence and a happy, productive and fulfilling life—something that we all want to achieve in our lifetime?

For both parents and educators, if you take away nothing else from this, I implore you to

understand how very limited your time in school is with your student with Angelman syndrome. As educators, you have this year, maybe the next year—but the families have the next 40, 50 or 60 or more years with their child with Angelman syndrome. Have a sense of urgency. We can't afford another week, two weeks, a month, a semester, or next year to get something in place for our kids. There are no 'mulligans' or do-overs—now is the time.

If the goal of education is truly for our children to learn then we all—parents, educators, administration and support teams—need to come openly and willingly to the table, ready to listen and work together to put forward the best plan of action for our students. Throw out pre-conceived notions and personal biases of what we believe is needed and truly work together to define and implement a learning environment where the student can and will learn and succeed. Stop trying to put our students in a pre-defined program or box, whether it is by virtue of the diagnosis of Angelman syndrome, their genotype, or any other preconceived notion of what our students will or won't achieve, or what they are or are not capable of doing. They are, first and foremost, people who are very capable of learning, growing and acquiring skills; they are not defined by their diagnosis. See them first as a person.

We need to have our children coming out of school with as much knowledge, skills and independence, not dependence, as possible. People with Angelman syndrome are incredible communicators—they are communicating constantly and we need to be truly willing to 'listen' and acknowledge their communicative intents. Learn to read their body language, intonations and vocal inflections. Learn their signs and gestures. Acknowledge their ability and use of technology, whether or not their eye gaze, tracking, finger isolation or swipe is perfect. Throw out the blind notion that people with Angelman syndrome don't have the prerequisite skills to use technology, learn, communicate or participate and be open to the perspective and knowledge that they can and they will. People with Angelman syndrome are absolutely capable of learning, far more than was previously recognized, thought or supported. Hold the bar of expectations high for your students and watch them rise to meet or exceed those expectations.

We had decided to repaint Kaitlin's bedroom and I knew Kaitlin had long, long ago mastered colors. I wanted her to have a choice in what color her room would be painted, so I made a wonderful color chart with nine bright color samples on the chart. As Kaitlin and I sat on her bed I told Kaitlin we were going to paint her room a new color and wanted her to choose what color she wanted. We already knew that purple was Kaitlin's favorite color but didn't want to assume that was the color she would choose. Showing the color chart to Kaitlin, I asked her what color we should paint her room, which she promptly vocalized 'puh-puh', her word for purple. She hadn't even looked at that fabulous color chart, so this couldn't be what she really meant! So I showed her the chart and asked her again and without hesitation she again vocalized 'puh-puh'. I needed to be 'sure' this was what she really wanted so, for the third

time, gave her the color chart and asked her again what color she wanted her room painted. She looked—or rather glared—at me, snatched the color chart out of my hand, said 'puh-puh', pointed to purple, then promptly tore the color chart in half and threw it at me.

How many times did I, did her team, and do we all, repeatedly ask people with Angelman syndrome to repeat over and over what they have already mastered, what they have already communicated? How incredibly frustrating for Kaitlin and for others with Angelman syndrome.

Communicate and collaborate with parents and previous team members. If we are considering teaching a task or skill, determine first if the person has already learned it and if they have then let's move on. We don't need to force people to demonstrate every single skill in order for us to accept that they have already learned it. How incredibly frustrating and humiliating it would be to have to demonstrate those tasks and skills which were previously mastered. Know your students, talk to the team and previous team members, read their records and previous IEP's, talk with and appreciate the student's parents by valuing their input and including them in the development of the IEP goals for their child.

People with Angelman syndrome have an innate ability to read others' intentions and true feelings toward them. If you are genuine and have their needs and success in mind, if you respect them and have set expectations high for them, they will work hard and be motivated to achieve. Please don't patronize people with Angelman syndrome, for this is thinly veiled. Don't enable your person with Angelman syndrome for this fosters dependence rather than independence. Show your respect for your student and person with Angelman syndrome by talking with them, not at them and certainly not about them, in age-appropriate language.

It is the hope and dream we have for all of our children—that Kaitlin, and her siblings, grow to achieve the greatest independence, happiness and success that they can and are able to achieve. We hope and dream they live a good life, surrounded by family, by friends of their own choosing, and that they understand, recognize and are thankful for the many talents, gifts and blessings bestowed on us all. As their parent, I want simply to love them, to teach them what I can, to give them a firm foundation of faith on which to base their lives and to help them find the opportunities to grow, mature and live an independent and happy life. It will look different for each of our children—as it should. Most importantly I want them to feel and know that they are all valued and, above all, loved.

[1] Eileen Braun has served as Executive Director of the Angelman Syndrome Foundation (ASF) in the U.S.A. since March 2003. Eileen has been instrumental in helping the ASF raise and fund over $8 million for scientific and clinical research, as well as helped many hundreds of families of people with Angelman syndrome.

Dare to Dream

Amy S. Girouard[1]

Hope and promise were two words that I never would have used when my daughter Ally was diagnosed in 2007 with Angelman Syndrome. She was not quite a year old and I thought her life was over before it had started. Ally has what is called deletion + for Angelman Syndrome which means that a large part of chromosome 15 is missing from her maternal chromosome. This affects gross motor skills, fine motor skills and speech. It essentially means that for Ally, everyday tasks like getting dressed, eating on her own and even walking are much more difficult and require more work on her part for her body to do what she wants to do.

At the time of her diagnosis, I saw a bleak future with minimal education, minimal interaction with the world around her and no chance for a typical future. Our hopes and dreams were completely shattered. I was terrified of what I read about Angelman Syndrome. My baby went from being my daughter to a set of symptoms and a diagnosis that I didn't understand. I had dreams of my daughter going to gymnastics, princess parties, sleepovers and a life full of promise and wonder. The outlook for my daughter was bleak at best. She had a potential for seizures, she would never be able to speak, she may or may not walk and she would always need 24/7 care. I couldn't begin to imagine that she could ever do a somersault, make friends, or even obtain an education. A life of promise and hope no longer existed for her.

I spent hours researching AS and found many articles that were depressing and seemingly outdated. I did, however come across one name that changed the way that I viewed my

daughter, her future and ultimately changed our lives forever. I found Dr. Calculator's name through an Angelman Listerv and investigated his work. For the first time, I had hope for Ally's future. Dr. Calculator came to my house when Ally was still under two years of age and talked with me at my kitchen table. He helped me to see her enormous strengths through the diagnosis. As we continued to discuss Ally, it was apparent that I misjudged her. I believed all of the negative literature about AS. I began to see her laugh and smile as a way to engage people into her world instead of a hallmark of a syndrome. I saw that she was communicating with us even without words. Her eyes, her hands, her body language were all communicating her needs to us. I had to open my eyes and my mind.

From then on, I slowly started to see her as my daughter again. I saw a different future than what I had imagined. That shift in my paradigm of thought was more profound than I could have ever imagined. When Dr. Calculator had told me that I needed to have "High Expectations" for Ally. I initially had no idea what that meant for her. I thought that high expectations were only reserved for people without any disabilities but most certainly didn't apply to people with AS.

Slowly, as the fog lifted, I began to come across stories of people with AS who have defied the expectations and the literature. I started to hear about how other children were walking and not only understood the world around them but had friendships and were fully included in school. I looked at what the common denominator was in all of these examples. It was twofold: The people with AS had full seizure control and were, for the most part, medically stable. The other was the profound paradigm shift of "High Expectations".

Being medically stable for Ally was a complex process. This is much more important than I ever realized. Medically stable meant that Ally was not having seizures and that her GI symptoms were under control. It takes time, patience and the right doctors to be able to ensure that your child is medically stable. No one on your child's team should assume that every medical issue your child is exhibiting is "just a part of AS."

Ally is in a small percentage of people with AS who do not have seizures. However, Ally does have an uncommon diagnosis of Eosinophilic Esophagitis. This is an autoimmune disorder where Ally has many allergies to food including gluten. This has caused her to have reflux, constipation, sleep disturbances, increased behavior such as hair pulling, aggression and what I call a "cloudy" brain. These all have been written in literature as typical Angelman behaviors and it is a common belief that there is nothing to be done about them. However we had the good fortune of having doctors and therapists on our team that wanted to know the reason behind the behaviors.

Instead of viewing maladaptive behavior as the norm, we began to look at her behavior as her

inability to communicate pain, discomfort, dissatisfaction or even anger that are all normal human emotions. We kept asking the question of "Why?"

Traditional testing showed nothing and Ally's behaviors; reflux and constipation seemed to be getting worse. Traditional medicine was able to diagnose Ally but could not help beyond the diagnosis. We turned to naturopathic testing to determine the allergens since we did not have many answers through traditional medicine. Once we removed the allergens, Ally's affect and demeanor began to brighten and she was lifted out of her cloud. For her, the special diet is a corner stone of her wellness and lays the foundation for her ability to learn, comprehend information, to work in therapies, strengthen all her muscles and communicate with the outside world. For us, it was an important lesson in looking at Ally as an individual and not the diagnosis. We carry this philosophy in other aspects of her life as well.

In conjunction with understanding Ally's medical issues, we needed to understand how to support Ally in other aspects of her life. In doing that we sought out therapists, teachers and support staff that viewed Ally as a child and not as a diagnosis of Angelman syndrome. When you look at your child as an individual and not as a diagnosis, the world seems to open up more than you can imagine. You will find opportunities to overcome challenges in a way that is best for your child and your family. You find "experts" who will provide a service whether it is medical, therapeutic or educational and follow your child's lead instead of following a prescribed set of rules and expectations. This has been the one of the most powerful paradigm shifts in how we approach Ally and what she needs to be successful.

AS affects Ally's ability to do many things that other children do with ease. She was five before she walked and at eight years old she still requires help to get dressed. Ally, who is considered "nonverbal", uses a communication device to let us know what she needs and wants. This device has the ability to grow with her so that she can communicate authentic thoughts. Ally understands what is being said to her, however, her ability to engage in reciprocal communication is still limited, which often causes her intelligence to be questioned. This is one of the more frustrating aspects of AS. Ally is intelligent and loves to learn. She loves to be one of the crowd and is proud to be an active participant in the classroom.

Our focus on Ally's education has been an evolution as well. When Ally was in developmental preschool, I knew that I wanted "high expectations" of her but I did not know what that meant. I knew that I wanted to believe that she could learn but I didn't know how to make that possible. Ally could not physically keep up with her peers, she babbled instead of talked; she did not even have a finger point. If one just looked at Ally, you would label her as "mentally deficient" and she was even assessed at four years of age to be functioning intellectually at an 18 months level.

I knew that the educators did not fully see Ally's potential but I did not know why. One day, I went to school during her therapy time and what I saw was the saddest but most enlightening moment of her education. A well-meaning therapist had Ally in a back room sitting at a table. The therapist was sitting across from her. The therapist had an iPad and on the iPad were pictures of objects. The therapist had those same objects in her hand. She repeatedly asked Ally to point at the representation of the object on the iPad.

Ally was compliant for a couple of trials and then put her head on the desk. To the therapist, this meant that she clearly had a lack of understanding of the material. To me, the opposite was true. Ally was not motivated by this kind of teaching. She is socially motivated and needs to understand the meaning behind the request. She does not do well when asked to "comply." It was a confusing and frustrating time for the school and us and it caused some friction between us. We knew that Ally comprehended more than we could ascertain, but we had no idea how to motivate her so that others could "see" the true Ally. It was then that we asked Dr. Calculator to re- assess Ally and her classroom for the following year.

Dr. Calculator's advice and unique perspective changed the way that we viewed Ally's education and enhanced kindergarten. The concept of full inclusion and what that meant was transformational. The kindergarten school team believed in this concept and Dr. Calculator found ways to refine it and helped the team capture Ally's strengths in learning. Since that time, we have advocated for full inclusion and immersed Ally in the community at school.

Currently, Ally is in second grade where she is fully included in the general education setting. This means that she receives physical, occupational and speech therapy in the classroom and she is not pulled out in order to receive those services. The team has found natural ways to incorporate therapy. For instance, physical therapy is incorporated in gym class and when she has to use the stairs in the school. Occupational therapy is provided while Ally is in art class and speech is provided while Ally is in reading group. She has the proper supports to help her succeed in the classroom.

Ally is allowed to make mistakes and learn from them. At school Ally is learning about addition, subtraction, spelling, reading and friendships. She is a unique learner and we are always looking at different ways of enhancing her educational experience. This is often challenging because it requires all of us to think outside the box and traditional education. It also requires different ways in which we can measure what she is learning. They have developed portfolios of her work and are videotaping her "writing" stories with her AAC device. They also are continually assessing what motivates her to learn and what we are finding is that her peers are motivating to her. She participates in morning meeting, is asked to identify wall words and uses the iPad to engage her in subjects that she has less motivation with such as math.

Perhaps the most heartwarming part of being included in school is the friendships that have happened. Ally now has friends who worry if she is not in school and who celebrate when she is there. Ally's school fosters a true sense of community for Ally and we are forever grateful for that gift. This experience has given us hope for Ally's future. Ally participates in jump rope after school and other community activities such as gymnastics. She has had a mixture of school and outside therapy but we also take as many opportunities as possible to integrate her into the community. This means we limit the outside therapies and we try to maximize typical activities. She requires extra physical help, which is sometimes difficult to obtain. However, we continue to push the envelope with educational and community inclusion which opens up more doors to opportunities and friendships. We hope that she will continue to learn from the world around her and will have teachers, aides and friends who believe in her potential. We hope that Ally will have the supports that she needs to be included both in school and in her community.

The next step in full inclusion for her education is to ensure that she is literate. Reciprocal communication continues to be a challenge. While we believe that her AAC device is the best for her right now, it still has limitations. The language system is complex and it requires everyone on her team to be well versed in it. Her educational team has been creative in understanding what she may know as far as spelling and math, however there is still a gap in what Ally wants to tell us about her thoughts, her day, her friends and the world around her. This leads to the question as to whether or not Ally can be taught to read and write. The answer is a resounding YES.

At first glance, given her challenges one might presume that Ally will not be able to read or write. She has limited communication so how will anyone be able to decipher what she knows and whether or not she has a unique thought on the subject? As we look toward the higher grades, this becomes so much more important and seems to be the next frontier in our journey. It is one thing to identify colors, spell simple words and add or subtract. What happens when the teacher requests that the students respond to a story with their own thoughts? How will she be able to still be fully included and fully participate in the classroom?

This is where we will have to become ever so creative in how we approach the problem. We are confident that she has her own thoughts and that she so desperately wants to communicate them. You can tell by the way that she grabs her crayons and will "draw" over and over again with great determination. You can see it in her eyes when she grabs a book and points to a character over and over again while looking directly at you. It is those times that we feel inadequate because she so desperately wants to communicate and her AAC device simply is not enough.

The search for a way for her to communicate with us will be a longer process. She has natural gestures that she uses quite successfully to get what she wants. She hands us the remote, she will take our hands and put them on an object to communicate what she wants from us. However that is simply communicating basic needs. This will be a difficult journey for all of us but one we are willing to take.

Our roles with Ally change and grow as all of us change and grow. Ally will always need a strong support system and we feel that it is our role to advocate for and build that support system to be there long after we are gone. As difficult as it sometimes is, we need to continue to push for those beautiful, normal, every day experiences like going to museums, play places, amusement parks and getting together with friends at the playground. It would be a disservice to her if we put her in a bubble and did not let her experience these things. Her quality of life is just as important as her therapies or education. We can never forget that and always advocate for her highest quality of life to experience normal life.

When you have a child with AS, it is easy to forget that there is life outside of Angelman syndrome. This is especially important in self-care as well as your intimate relationships. It is crucial to make time for yourself, whether it is a 10-minute rest period, exercise, a walk or a bath. It is not always easy, but taking care of yourself is essential to making sure that your children have the best life possible. Taking care of your intimate relationships is another important lesson that we have learned. My husband and I still have dates, overnights and celebrations of us as a couple. It is important to nurture that relationship because it has given me strength when those dark days of insecurity, grief and exhaustion come around. It is important to acknowledge those dark days and let yourself feel the emotions. This was not the life we had planned, however we have to make the best of each and every day. Having fun as a couple will pave the way for having fun as a family. This will provide you with some normalcy that is much needed.

Looking back there are always things that you wish you could have done in a different way. I wish we worried less, laughed more and been present in the moment. As an example, we took a weeklong trip to an amusement park and I was so worried about seizures that I covered her eyes through most of the rides. I had to always remind myself that she is first and foremost a little girl who loves roller coasters, being around people and wonderful magical characters. I needed to remember to put myself in her shoes, follow her lead and let her be a child all the while keeping her safe and not in harm's way. It is a lesson that I am constantly being reminded of when we are out with her. If we are to give her the best life possible, we need to widen the parameters, teach her safety awareness and strike that balance of safety and fun.

It is also important whenever possible to hold your child with AS to the same values and standards as other children in the house. This admittedly is easier said than done and we are

still working on it. We have a 13-year-old son, Josh, as well as Ally. He is a gift in our lives and they have a truly special relationship. He is protective of her and, at the same time, he teaches her to be a kid. He thinks of things that we would not such as teaching Ally how to play rock, paper, scissors; wheelbarrow; and tag. He treats her like a big brother normally would and with that comes great rewards. It also means that he questions why Ally is not doing the dishes or picking up her toys, as he must do. It is difficult to maintain this because Ally needs that extra help to do these tasks. However, we need to always ensure that Ally knows that she is a member of the house as well and that she needs to do chores too. This is a work in progress and may always be a challenge. However, what that does is even the playing field a little between siblings and it gives Ally a sense of purpose and belonging in the house. Everyone, no matter what your age, color, race, creed or disability needs a sense of purpose. This is a value that we instill in my stepson, Matt as well. He is a crucial part of our family and will promote them long after we have gone. I hope that we always strive to achieve this goal with our family.

We have had the good fortune of having many professionals who see Ally as a little girl who happens to have Angelman syndrome. The best professionals will give their advice based upon experience and education, however they also respect us as the parents who ultimately know Ally the best. They allow us to make our own decisions based upon their recommendations and our personal values. If those conflict, they respect our personal values while providing us the education and advice. They never assume that something is "just part of AS"; they want to know why something is happening and to ameliorate it if possible. The best way that professionals can support people with Angelman Syndrome and their families is to take a true person/family directed approach. We need professionals for advice and assistance in many areas. We need your support. We also need professionals to respect our values and trust us to make the best decisions possible for our family, provided that we have all of the information to do so. We expect that every professional will have high expectations for her and to always encourage us to shoot for the stars.

Ultimately, we wish for Ally a sense of purpose, normalcy, friendships and competence. We know that she will always face enormous challenges in life and we hope to ease that burden every day. This story has yet to be written. There are so many possibilities for her that we may not even know right now. The best that we can do is to keep looking for opportunities that challenge her and us for a better future. It will not be easy and there will be mistakes made. Those mistakes, however, will lead us to new opportunities and a fuller future for all of us. I want to know Ally's thoughts about life and what are her hopes and dreams. I want her to know that anything is possible and that she is not defined by AS. The future is brighter now than it ever has been for people with Angelman Syndrome. That flame will only grow brighter as more people believe in the potential of people with Angelman Syndrome.

Families, communities, schools and governmental agencies need to work together to break down the barriers to a full life. Ally and others like her have so much to offer. We believe that Ally and others with AS have so much untapped potential. If all of us work together we can create a firestorm of hope and promise. We promise to always let Ally's light shine and work to break down the barriers for all people with Angelman Syndrome so that everyone can have the promise of a future that is filled with every day experiences, friendships, and hope.

Acknowledgments

I would like to dedicate this prologue to my husband Chuck Crush. Together we have created a family of hope, love, and a belief for all of our children that anything is possible. You are my inspiration.

[1] Amy Girouard, MSW, LICSW and her husband Chuck are volunteers for The Foundation for Angelman Syndrome Therapeutics (FAST), promoting awareness as well as organizing fundraising events. Amy also serves as a disability rights advocate.

Training for a Marathon: Raising an Exceptional Kid Who Happens to Have Angelman Syndrome

Susan Ravellette[1] - Ryan's Mom

I will never forget the evening that I received the "official" call that Ryan had Angelman Syndrome (AS). I had suspected that Ryan had AS for several months due to hours and hours of Internet research trying to find an answer to his developmental delays. However, I did not tell anyone of my suspicions, not even my husband. So when that call came that evening from Ryan's neurologist, it dashed every hope that I had that Ryan was simply a little slow in reaching milestones. What this call did was to confirm what I already knew but did not want

to admit. We were fortunate that the news was delivered by a wonderfully empathetic neurologist who called me at home that evening so as not to torture us by making us come into her office the following day to deliver the news that Ryan had a mutation of the UBE3a gene. Right after I hung up the phone, my father-in- law happened to call and I blurted out the news to him before I had told anyone else, not even my husband. The first thing he said to me was "Ryan is still the same Ryan that we all love." He was right, but it took a while to get there. It was very difficult news to hear and process.

Ryan is now almost thirteen years old but he was only nineteen months when he was diagnosed with the UBE3a genotype, one of the rarer forms of AS. The information on AS in general was so limited at that time and there was virtually no accessible information on Ryan's specific genotype. I did realize right away, because of my earlier research, how severe this syndrome was portrayed. Because I had not shared my earlier suspicions with my husband; the diagnosis blindsided him. We emailed a lot of friends and family that evening to tell them the news but said we just could not talk about it yet. It was a very difficult time and one that still makes both of us emotional when we talk about it. It is one of the most heartbreaking events for any parent and one you cannot prepare yourself for. For at least a year following Ryan's diagnosis, I cried nearly every day. But as I tell every newly diagnosed parent I speak with, it gets much better. It is a little sad to me, looking back on those earlier years, that all we thought about was AS all the time. Now I see Ryan as much more complex and interesting, and the last thing I think of when I think of Ryan is AS.

It is hard to remember what our dreams were for Ryan when he was that young. I think we believed he would grow up like we did, playing little league, having friends over to play, going to college, and hopefully graduate school. We started the college fund when he was born and went about our lives like all our other friends with kids until we started getting the warning signs. Then our life became inundated with various therapies, agencies and paperwork.

Because the information was so limited, all I really knew was that the current literature stated that one hundred percent of kids with the more common form of AS, deletion positive, would have onset of seizures by age three. That fact absolutely terrified us. We constantly watched Ryan for any signs of seizures, which was incredibly stressful. It was also frustrating because there was no information on what the likelihood of seizures would be in the case of a person with the UBE3a mutation genotype. We were scared about the future and, because Ryan is nonverbal, having to trust that the therapists and teachers who would be working with him would keep him safe was a constant worry. It still is. We worried about Ryan being provided for after we were gone and spent a lot of time estate planning. Just a few months after his diagnosis, we learned that I was a carrier of AS and therefore had a fifty percent chance of having a child with AS. That news was nearly as devastating as the initial news of Ryan's

diagnosis.

When you decide to have a family, you never imagine that you are going to have a child who will face huge physical and cognitive challenges. Our whole family dynamic seems so drastically different from my friends who have children without such serious issues. But that said, I cannot imagine a life without Ryan as he is, not who I thought he would be before he was born. However, having Ryan did bring some isolation to our lives, much of it self-imposed. There are the friends and family who have no idea how to react to your news. As a working parent, I happened to be pregnant at the same time as three other co-workers. We all had boys born just a few months apart. When we were given the diagnosis, I was not able to be around any of them or their children for quite some time because it was too painful to watch the life that Ryan should have had. The life that I envisioned as a parent was gone. Birthday parties were my Achilles heel because it was so difficult to watch the celebration of a child who could run, jump, talk and do so many things that Ryan was struggling to learn and might never be able to do. Birthday parties also felt isolating because Ryan would need me to help him participate in the party activities while all the other parents would be off to the side socializing.

It became easier to isolate ourselves from our friends and family and to seek out support from other Angelman parents who were going through the same thing. One Angelman parent and I became very close and I would often call her, crying on the phone, because no one would understand. I felt that in front of co-workers, friends and family, we had to stay strong so people would not just pity us. I would find excuses to get out of social gatherings with friends who had kids that were Ryan's age because I just could not listen to parents talking about their children's lives that seemed so easy compared to the life we had with Ryan. How nice it must be that your child can learn to walk without hours, weeks and months of physical therapy? I just could not listen to it. However, now I feel quite the opposite. I love to brag about Ryan and his achievements. I realize all children have obstacles and challenges. I think Ryan would be a little offended if he knew that I was, at one time, sad when comparing him to other children. Now, I think of all the amazing things he is capable of and how much people enjoy his company. The upside is also not seeing the "tween" attitudes so many of my friends' kids are showing these days.

As time went by I made a decision to be an active member of the Angelman community, including serving on the Board of Directors for the Angelman Syndrome Foundation. We found a wide variety of AS families that were living their lives in a way that gave us hope. I remember the first house we went to of a family with a daughter that was a few years older than Ryan and finding that it was a "normal" house. That was such a relief to know that we could stay in our house and do modifications to make it safe for Ryan without turning it into an institution. We started fund-raising to find treatments for AS. We tried to become as

involved as possible so that we would have access to what was happening in the world of science and education for AS. We became fierce advocates for Ryan, both for his medical treatments and his introduction into the public school system.

When we first placed Ryan in the public school system, the only program offered was a special day class for three-year-olds. Until that time, Ryan had been in "typical" day care. When he started preschool with the school district at age three, he was placed in a class that had children who were highly autistic. Ryan was the polar opposite of an autistic child. He was engaging, happy, full of laughter and curious. It worried us that it would inhibit his social development. We decided to have him spend half of his day in this special day class but continue to spend half of his day in a typical daycare preschool setting.

When it came time for Ryan to start kindergarten, we were adamant that he attends his neighborhood school in a fully included setting. How to go about truly doing that? We had no idea. The District really tried to convince us to send Ryan far outside our neighborhood to a school that had a special mild to moderate day class. I made a point of meeting the teacher from this class, who upon learning Ryan had AS, said to me "if he exhibits 'typical' Angelman behaviors, he does not belong in this class." That is the first moment I realized that many people, especially in the educational field, would see the diagnosis first and my bright, funny, beautiful child second. In my mind, Ryan was still Ryan who happens to have AS.

While our neighborhood school did not have a special education teacher on staff, we were fortunate that the school embraced Ryan. The principal was exceptional and let it be known that Ryan had the right to attend that school as much as any other kid in the neighborhood. However, it soon became apparent that none of us really had a clue on how to successfully include Ryan. It was difficult to figure out how to incorporate his IEP goals into day-to-day classroom activities. From our viewpoint though, we truly believed that Ryan learned better from being around "typical" peers than he did from being segregated in a special education classroom. Over time, the school built a support system for kids like Ryan including hiring a special education teacher to help "oversee" the curriculum of the kids with AS. I am proud that Ryan was the trailblazer at his school, even if he was not aware that was his role. Ryan was the first with his level of needs at his elementary school, but many other kids have enrolled at that school since due to the successful program that was built.

However, over time we have come to realize that the inclusion setting only works if all the educators are committed to engaging Ryan as one of their students. As he transitioned to middle school, it became clear that the teachers did not have the time, or the inclination, to truly educate Ryan. For the first time since kindergarten, Ryan now spends half of his time in a special education classroom and half the time in general education classes such as drama, student government and music. The time spent in general education, we came to realize, was

not very productive the older Ryan became. We honestly fought a lot harder earlier in Ryan's education but as time goes on, it is a real struggle to maintain that level of advocacy. The pressures of life and other children diminish the energy you have left to keep fighting. We know Ryan is capable of so much more than what he is given but it is a constant challenge to convince everyone else that he really is quite intelligent in his own way. He is constantly amazing us with random acts that show how high his cognition level is and how he is engaged in the world around him. Just recently, Ryan, my daughter and I were leaving to go to the library. My daughter was searching for her e-reader and as she and I were talking about where it may be, Ryan went to our living room couch, pulled aside a cushion, pulled out the e-reader and handed it to his sister. A small thing for some people but it was huge for us to show how much he really takes in what is happening around him.

Currently, Ryan is in the seventh grade. He actually seems to be enjoying school; he is especially fond of the large "smart board" in his classroom. Ryan is a tech geek and loves all things electronic. At home, he loves spending time with his sister Sarah, who is almost nine years old. He often searches for her around the house and just wants to be in her presence. His friendships have become more challenging as his peers enter their teen years. Often their interests do not necessarily coincide with Ryan's interests. We have seen some strong friendships with boys taper off while girls will actively seek him out. Although with girls their interactions come off at times more motherly and not a true peer to peer relationship. It also feels artificial to have to be the one, as the parent, to create these friendships in a way. But the only way that Ryan will spend time with his peers is if we make it happen. Again, I often think about the other kids and how they arrange their time spent with friends on their own and without parent involvement. It seems so much more natural. However, because Ryan has been fully included at school and attends many local summer camps, it is a rare outing in our community where someone does not say hello to Ryan. He knows a far greater number of people in our community than my husband, daughter, and me combined.

For Ryan to truly enjoy his life, we believe that comes from gaining much more independence. We are not unrealistic in that we know Ryan will require twenty-four hour supervision for the rest of his life barring any life changing medical treatments. We wish for him to live in a home with as much independence as possible. Both my husband and I wanted to live on our own when we became adults, and while Ryan has not voiced an opinion to us, it is hard to imagine he would think any differently. He should get to experience living away from his parents and having his own life.

At our first meeting with a geneticist after Ryan was formally diagnosed, the doctor said to us "Ryan would be severely mentally retarded and he would never live independently." As to the first portion of that opinion, we simply never believed that. As to the second portion, my thoughts are "who really does live independently?" I think it would be said that I have lived

independently ever since I was an adult, but that is not entirely true. If my drain gets plugged, I call a plumber to fix it. If my electricity goes out, I call an electrician. If a burglar breaks into my home, I call the police. I need other people to help maintain my "independent" lifestyle to stay comfortably in my home. That is as independent as needing someone to help ensure Ryan's safety.

We have seen how some siblings of kids with AS feel the pressure to be the care provider when the parents no longer can fill that role. We do not want that. We have always told our daughter that it is our responsibility to make sure that Ryan has adequate supports in his life and it will not be her job to be his primary caretaker. However, we do stress that it is her job, as his sister, to care what happens to him. We are fortunate that our daughter is wonderfully empathetic and we have no doubt that she will always be in his life. We also realize that we need to build a community of people that will look out for his well-being. We have to build this community so that he has purposeful social interaction. In the school environment that is rather built in but outside the school setting it is much more difficult to engage that level of social interaction.

When Ryan was just a little over one and entering the early intervention services, a nurse who was evaluating him told me that his life, and my life, was a marathon not a sprint. That has stuck with me all these years. You can get so caught up in those early years feeling like you are not doing enough, you should be doing more, you should be working with your child every waking second because you are afraid you will miss this magical "window" of development. It is not true. You can only be the parent who helps with the therapy, you are not the therapist. I would feel overwhelmed with the amount of therapies I was "supposed" to be doing with Ryan in between each session. I always felt guilty that I was not doing enough. Then it occurred to me that Ryan should be allowed to be a kid like any other kid. At the age of two or three and really to this day at the age of twelve, he deserves to have time to explore the world on his own without constantly being guided by adults around him. It turns out he is thriving and growing as long as we give him the right environment and let him figure things out without always jumping in to help him in every task. To be completely honest, the kid is a master of manipulating people into doing things for him with his movie star good looks and bright smile. He knows how to do a lot more than he generally shows he is capable of doing. One of the first things we tell a new care provider is "don't let him fool you."

I want the professionals that work with Ryan to see him as an individual first. It is so important to set high goals and not to lower the bar simply because it is easier. I want professionals to admit when they are unfamiliar with a program or regimen a child is on and work with the parents to find solutions. During his elementary school years, we fought the school district to fund an expensive, sophisticated AAC device. His ability to master it and perform some pretty sophisticated communicative skills with this device was impressive.

When he transferred to middle school, this was lost because the staff did not understand the device and never asked for or received the necessary training to implement the device correctly. As a result, Ryan no longer uses this device, but he does use an IPad for communication, albeit with a much lower level of complexity. We were forced to ultimately switch to the IPad because that is what the professionals at his new school were comfortable with and we had already lost valuable time in Ryan's communication evolution. Instead of looking at Ryan's needs and levels of ability, they were focused on their own limitations. They did not want to get fully trained because they believed they knew what was best for a child that they did not even know.

We dream that Ryan will have the life that he chooses to live. We dream that somehow he will be able to communicate those thoughts and desires. We dream that he has a full and real social life of his choosing. We dream that he will get a college education in whatever form that takes and find a job that can help financially support him. We hope that he continues to look at life with a glass half full approach that so many of us wish we could emulate.

[1] Susan served on the Angelman Syndrome Foundation Board of Directors from 2005-2014 and the Western Area Chapter Board of Directors from 2005-2008. She is a Pro Se Staff Attorney for the United States District Court in the Southern District of California.

List of Contributors

Caroline Ramsey Musselwhite	Assistive Technology and Emergent Literacy Consultant , Phoenix, AZ, Arizona
Charles A. Williams	Department of Pediatrics, Division of Genetics and Metabolism, University of Florida, Gainesville, FL, USA
Erin S. Sheldon	Angelman Syndrome Literacy Project, Kingston, Canada
Giulio E. Lancioni	Department of Neuroscience and Sense Organs, University of Bari, Bari, Italy
Hilary Johnson	Scope's Communication and Inclusion Resource Centre, La Trobe University, Victoria, Australia
Jane Summers	Psychologist and Behavior Expert, Centre for Addiction and Mental Health, Toronto, Ontario, Canada
Jeff Sigafoos	School of Educational Psychology and Pedagogy, Victoria University of Wellington, New Zealand
Jennifer M. Mueller	Pediatric Genetic Counselor, Division of Genetics and Metabolism, University of Florida, Gainesville, FL, USA
Mark F. O'Reilly	Department of Special Education, University of Texas, Austin, TX, USA
Peter B. Marschik	Department of Physiology and Neurolinguistics, Medical University of Graz, Graz, Austria Karolinska Institute, Stockholm, Sweden
Robert Didden	Behavioral Science Institute and School of Education, Radboud University, Nijmegen, The Netherlands
Scope Victoria	La Trobe University, Victoria, Australia
Stephen N. Calculator	Department of Communication Sciences and Disorders, University of New Hampshire, Durham, NH, USA
Teresa Iacono	La Trobe Rural Health School, La Trobe University, Victoria, Australia
Vanessa A. Green	School of Education, Victoria University of Wellington, New Zealand

CHAPTER 1

Clinical Features, Medical Issues, and Diagnostic Testing in Angelman Syndrome

Jennifer M. Mueller, Charles A. Williams*

Raymond C. Philips Unit, Division of Genetics and Metabolism, Department of Pediatrics, University of Florida College of Medicine, Gainesville, Florida

Abstract: This chapter reviews the clinical and medical aspects of Angelman syndrome (AS) and the genetic mechanisms that create a deficiency of the function of *UBE3A* on the maternally-derived number 15 chromosome. Genetic mechanisms causing AS include cytogenetic deletions, imprinting center defects, paternal uniparental disomy and *UBE3A* mutations. We review the clinical criteria for the diagnosis of AS and discuss genetic confirmatory testing. The chapter explores the natural history and characteristic features of AS including severe developmental delay, speech impairment, gait ataxia, microcephaly, seizures, and unique behaviors. Finally, we will discuss the implications for learning and overall function.

Keywords: Angelman syndrome, Behavior phenotype, Chromosome disorder, Chromosome 15, Developmental delay, Genetic imprinting, Intellectual deficiency, Neurogenetics, *UBE3A* gene.

INTRODUCTION

Angelman syndrome (AS) is a neurodevelopmental disorder caused by a lack of the ubiquitin-protein ligase protein, E6-AP, in the brain. AS is characterized by severe developmental delay and speech impairment, a unique behavior with a happy demeanour (includes smiling, excitability, and frequent laughing), small head size (microcephaly), seizures, and gait ataxia and/or tremulousness of the limbs. Although developmental delays may be seen around six months of age, the unique clinical features usually occur after the first year of life. Therefore, obtaining a timely diagnosis can prove challenging- especially since there may be

* **Corresponding author Charles A. Williams:** Division of Genetics and Metabolism, Department of Pediatrics, 1600 SW Archer Road, PO Box 100296, Gainesville, Florida 32610; Email: willicx@peds.ufl.edu.

Stephen N. Calculator (Ed)

no obvious dysmorphic features to help direct the physician. The distinctive behavioral features are crucial in suspecting a diagnosis but may not emerge until early childhood. This chapter will further explore the natural history, implications for learning and overall function, genetics and disease mechanisms, clinical testing, and gene therapies under investigation in AS.

CLINICAL FEATURES

Historical Overview

Angelman syndrome (AS) was first described in 1965 by English physician, Harry Angelman (Angelman, 1965). The three children in his report exhibited a stiff, jerky gait, absent speech, excessive laughter, and seizures. By the early 1980s, the first published case reports appeared in North America. In 1987, an Oregon Health Science Center physician, Ellen Magenis, identified children with microdeletions (or loss of genetic material) on chromosome 15 at the q11.2-13 region. At that time, deletions of this region were only associated with Prader-Willi syndrome (PWS). However, the children reported by Dr. Magenis showed severe developmental delays and seizures which are not expected in PWS. Investigators realized these children actually had microdeletions on the *maternally* derived chromosome 15, while individuals with PWS had deletions of the *paternally* derived chromosome 15. By 1991, further research determined that AS could also be caused by two copies of the paternal chromosome 15, as well as disruptions of the regulatory region (the Imprinting Center) in 1993. Finally in 1997, the AS gene, *UBE3A*, was isolated and researchers came to better understand its role in neural development.

Incidence

Cases of AS are seen throughout the world in individuals of all races. Although the true incidence remains unknown due to challenges of early identification, misdiagnosis, *etc.*, it is estimated to be 1 in 12,000 to 1 in 24,000 (Clayton-Smith & Pembrey, 1992; Steffenburg, Gillberg, Steffenburg, & Kyllerman, 1996; Mertz *et al.*, 2013).

Clinical Features

In AS, the prenatal history and development of the fetus is usually normal. At birth, babies with AS have a normal phenotype including birth weight and head circumference. Difficulty with sucking and feeding may be the first abnormality noted. By six months of age gross motor delays and muscle hypotonia may become noticeable, but the unique clinical features are usually not apparent until after one year of age. The initial diagnosis is often suspected in older infants based on a combination of absent speech, happy demeanor, and tremulous, jerky movements. However, a clinical diagnosis may still be missed for several years since children with AS do not have any significant craniofacial dysmorphism (Fig. **1.1**). The typical age of diagnosis is between two and five years of age. There are useful consensus criteria to establish a clinical diagnosis of AS (Williams *et al.*, 2006).

Fig. (1.1). Individuals with Angelman syndrome, proven by genetic testing.

Natural History

Newborns with AS can have low muscle tone, trouble initiating suck and sustaining breast and bottle-feeding. Varying degrees of mandibular prognathism

CHAPTER 2

Learning Characteristics of Students with Angelman Syndrome and Related Instructional Strategies

Erin S. Sheldon*

Angelman Literacy Project Kingston, Ontario, Canada

Abstract: Angelman Syndrome (AS) is associated with a constellation of disabilities. Students with AS present in the classroom with complex communication needs, cognitive disabilities, and differences in how information received through the senses is perceived and integrated. These students also have significant strengths: strong social motivation, a desire to communicate and interact, a positive affect, and strong attention when instruction is socially interactive and personally meaningful. Understanding the nature of the learning differences in AS is essential to planning how to meet these student's educational needs and successfully include them in academic instruction.

Keywords: Accommodation, Aided language modeling, Alternative assessment, Augmentative and alternative communication (AAC), Angelman syndrome, Apraxia, Assistive technology, Attention, Autism, Complex communication needs, Dyspraxia, Education, Emergent literacy, Engagement, Epilepsy, Fine motor, Gross motor, Sensory integration, Video modeling.

INTRODUCTION

This chapter is a description of the learning characteristics that are often observed in students with Angelman Syndrome (AS). Angelman Syndrome is associated with specific differences and disabilities that require accommodations in the classroom. Strategies to support and accommodate these differences to maximize learning opportunities are discussed. This material serves as a precursor to Chapter 9, Comprehensive Literacy Instruction for Students with Angelman Syndrome, where the issue of literacy instruction is examined in depth.

* **Corresponding author Erin S. Sheldon:** 37 Inverness Crescent, Kingston, ON, K7M 6P2, Canada; Tel: (613) 328-7632; E-mail: sheldon.erin@gmail.com.

Stephen N. Calculator (Ed)

My daughter Maggie is in the 6th grade. When Maggie was first diagnosed with AS as a toddler, we turned to the research literature to discover best practices and guide us to meet her educational needs. The research detailed the extent of developmental delays we should expect but offered minimal direction to support us to plan her education. We were left with unanswered questions: What did her AS mean for how she should be educated? What should we do differently as a result of her diagnosis? What educational strategies could help her access the instruction and learning in her classroom? How could we maximize her participation? How could we assess her learning and use that assessment to direct our interventions? I returned to graduate school and studied the learning characteristics of students with AS. This chapter is a summary of these characteristics, drawn from the literature and analyzed for the purpose of planning for how to meet the educational needs of our students.

LEARNING CHARACTERISTICS

Students with disabilities may differ from their peers who are developing typically in six broad areas: communication, cognition, physical abilities, senses, attention, and affect (Erickson & Koppenhaver, 2007). Students with AS are complex because they tend to have observable differences in all six of these areas. In addition, students with AS often have medical needs that impact their learning in the classroom. This section describes the differences that school teams might observe in these students as well as broad strategies to accommodate these differences.

Communication

Angelman Syndrome is associated with complex communication needs and minimal speech. The expressive communication disability is disproportionately more severe than the student's overall developmental delay, cognitive disability, and receptive understanding (Gentile *et al.*, 2010). These students require intensive support in school for communication skill development. What students with AS can express is the least of what they understand. Given the pervasive nature of their profound communication disability, what students with AS can express must be recognized as the floor, not the ceiling, of their comprehension.

Dyspraxia And Apraxia

Oral speech is rare in students with AS. Families often report spontaneous words and even sentences, yet these utterances can rarely be performed on demand and may never be repeated. This inability to produce or imitate speech on command is consistent with the nature of apraxia (difficulty initiating a motor response) and dyspraxia (difficulty planning and executing a motor response). Penner *et al.* (1993) documented significant motor planning deficits in adults with AS and characterized these deficits as a global developmental dyspraxia with particularly severe oral motor impacts.

Many students with AS develop some manual sign language, but their signs tend to be recognizable only to familiar people within familiar contexts (Calculator, 2013). Manual sign language development is complicated in students with AS for several reasons: apraxia inhibits the initiation of motor movements, dyspraxia causes difficulty planning and coordinating motor movements, ataxia results in tremulous movements and low muscle tone, and a generalized delay in fine motor skills contributes to poor accuracy in signing. The manual signing abilities of students with AS are unlikely to meet the student's needs enough to be understood by unfamiliar people, relate thoughts on abstract concepts, share personal stories, or report on their learning and comprehension. Educators need to plan for multimodal strategies to support communication. These multimodal strategies include manual signing, natural gestures, Enhanced Natural Gestures, vocalizations, graphic symbol displays, speech generating devices (SGDs) and other forms of alternative and augmentative communication (AAC). Fig. (**2.1**) shows students with AS using both light-tech paper-based AAC systems and high-tech speech generating devices.

Dyspraxia inhibits the ability to implement and enact what would normally be volitional or voluntary movements (Donnellan, Hill, & Leary, 2013). Dyspraxia is observed when students accidentally activate the wrong limb to move, seem to struggle with how to position their body in space, appear to freeze in space and then suddenly engage in a poorly targeted movement, or appear to ignore prompts to respond with a motor movement. Students who struggle with these kinesthetic dilemmas (Bauman & Kemper, 1994) may be good candidates for special

CHAPTER 3

Broad Considerations Driving AAC and Related Instruction for Individuals With Angelman Syndrome

Stephen N. Calculator*

Department of Communication Sciences and Disorders, University of New Hampshire, Durham, NH, USA

Abstract: Augmentative and Alternative Communication (AAC) and related assessment and intervention practices for individuals with AS might be best viewed as fluid, reciprocal processes that are important to the extent that they contribute to individuals' patterns of participation and inclusion in valued events. This is true irrespective of etiology of AS, severity of challenges, or educational placement. Educators, speech-language pathologists, and others are encouraged to emphasize practices that foster individuals' independence, autonomy, and quality of life. The use of standardized tests is questioned from this perspective. More viable, ecologically valid alternatives are presented. Several assessment and intervention procedures related to this construct are discussed.

Keywords: AAC, Angelman Syndrome, Alternative communication, Assessment, Augmentative, Disability, Discrepancy analysis, Ecological validity, Education, Generalization, Genetics, Handicap, Intellectual, Intervention, Matrix, Regression, Skill clusters, Self-determination, Standardization.

INTRODUCTION

Chapters 5 (Fostering Communication Skills in Beginning Communicators) and 6 (Fostering Effective Communication Skills in Individuals With Angelman Syndrome Who Rely on Symbolic Methods of Communication) of this book include discussions of practices that may be useful in facilitating effective

* **Corresponding author Stephen N. Calculator:** University of New Hampshire, Department of Communication Sciences and Disorders, Hewitt Hall, Durham, NH 03824, USA; Tel: (603) 862-3836; Fax: (603) 862-4511; E-mail: Stephen.calculator@unh.edu.

communication skills in beginning communicators, and those exhibiting more symbolic communication skills, respectively. However, regardless of individuals' current communication abilities and their potential to acquire more complex skills, a common set of considerations may come into play. These principles look at assessment and intervention as continuous and fluid processes. In other words, rather than considering assessment as a precursor to intervention, the two are viewed concurrently, with each exerting impact on the other in terms of the process of decision-making. As such, strategies that apply to assessment have concurrent implications for intervention as well. These are the foci of this chapter.

Avoiding One-Stop Shopping

Current practices often amount to speech-language pathologists (SLPs), educators, and others gathering snapshots of individuals' performances at points frozen in time. As difficult as it is to administer formal, standardized tests to individuals with AS, professionals often attempt to do so. Testing is often scheduled over one or several days, with a battery of procedures administered. Individuals' linguistic, cognitive, sensory, motor and other challenges call for systematic modifications of testing protocols in an effort to yield valid and useful results. However with or without modifications, over-reliance on these tests can lead to treatment decisions that are both ill conceived and ecologically invalid.

Ecological Validity

For one, content evaluated through administration of formal standardized tests often bears no resemblance or significance in relation to the types of communication demands and expectations individuals face daily. As such, the results of such tests often tell us little about what constitute significant problems and how their remediation might lead to more functional skills in everyday life.

If deficits identified by such tests are subsequent targets of intervention, what can we expect in terms of changes in individuals' levels of participation, interaction, and inclusion? How does an individual's inability to demonstrate comprehension of vocabulary or specific syntactic structures on a standardized test relate to misunderstandings during conversations at home, school, work, and elsewhere in the community? If individuals are to participate optimally in academic, work, and

other activities, it is important that they understand the language associated with them. To what extent is such content reflected in standardized tests?

Taking this one step further, it may make better sense to pre-test a child's language abilities if full participation in classroom activities is expected. Looking specifically at vocabulary, for example, words subsequently taught to individuals are those very words they are expected to encounter throughout their day. Assessment leads directly to intervention. Once involved in the corresponding classroom and other activities, what evidence do we have individuals 'know' the vocabulary they have been taught? This may be determined by examining participation patterns in the classroom and other settings – intervention leads to assessment. By avoiding one stop shopping, SLPs and others are mindful of intrinsic limitations of these tests. At the same time they recognize the value of other procedures that are better aligned with functional skills and outcomes.

Some Pitfalls of Standardized Testing

When team members opt to use standardized tests, procedures may not be implemented in a standardized manner. To the contrary, the adept professional recognizes the need to break standardization in order to obtain a more valid picture of individuals' true skills. It is important to remember that once we break standardization, normative data can no longer be used. That said, there are many instances in which team members report developmental ages, quotients, percentiles, z scores, stanines and other statistical measures despite their having administered the corresponding tests in a non-standardized manner.

A true danger emerges when test results are translated into developmental ages. For example, it is common to hear a SLP disclose an individual's performance placed him in the 18-24 month range for receptive language. Similarly, based on a standardized, though modified, test of intelligence, a specialist might suggest an individual's performance is consistent with a mental age of 3 years; 0 months.

One may question the meaning of general descriptive measures such as these. The 16 year old adolescent with AS who achieves these scores certainly presents a very different profile of strengths and weaknesses, learning style, functional skills, and needs than a typically developing younger child of 18-24 months or 3 years of

CHAPTER 4

Overview of Augmentative and Alternative Communication and Special Considerations for Individuals With Angelman Syndrome

Stephen N. Calculator*

Department of Communication Sciences and Disorders, University of New Hampshire, Durham, NH, USA

Abstract: The primary purpose of this chapter is to provide readers an overview of augmentative and alternative communication (AAC) as a prelude to subsequent chapters exploring this topic in greater depth. Basic material pertaining to assessment and intervention practices and principles is presented. The chapter begins by differentiating the two applications of AAC as augmentations of or alternatives to individuals' existing methods of communication. Information is provided regarding the impact of AAC systems on speech. Next, the four components of AAC systems; (a) symbols, both aided and unaided; (b) techniques, which include direct selection and scanning; (c) devices and other aided and unaided methods; and (d) strategies are reviewed. Educators and others are encouraged to address all four of these components when designing and implementing AAC programs that consider individuals' current and future communication needs.

Keywords: AAC, Alternative, Angelman Syndrome, Assessment, Augmentative, Communication, Devices, Education, Genetics, Intellectual, Intervention, Severe, Speech, Strategies, Symbols, Techniques, Technology.

INTRODUCTION

The purpose of this chapter is to provide an overview of augmentative and alternative communication (AAC) by discussing general principles related to assessment and intervention. This material will serve as a springboard for content covered in subsequent chapters. We begin by discussing the two different

* **Corresponding author Stephen N. Calculator:** University of New Hampshire, Department of Communication Sciences and Disorders, Hewitt Hall, Durham, NH 03824, USA; Tel: (603) 862-3836; Fax: (603) 862-4511; E-mail: Stephen.calculator@unh.edu.

applications of AAC, one as an augmentation of and the other as an alternative to individuals' existing methods of communication. The former includes a description of the impact AAC may play on individuals' development of speech and discussion of the importance of using AAC to complement individuals' extant methods of communication. Next, the use of AAC as an alternative method of communication is discussed primarily in relation to its role in reducing occurrences of challenging, or, socially inappropriate behaviors. This subject is addressed in much greater detail in chapters 5, Fostering Communication Skills in Beginning Communicators, by Jeff Sigafoos and his colleagues, and eight, Application of Principles of Applied Behavior Analysis in Addressing Challenging Behaviors of Individuals With Angelman Syndrome, by Jane Summers. We then review the four basic components of AAC systems: (a) symbols, (b) techniques, (c) devices and other aided and unaided methods, and (d) strategies. The content leads to a conclusion that AAC is a viable option that should be considered for all individuals with AS.

AAC APPLICATIONS

As the term suggests, the two basic purposes of AAC relate to its uses as augmentation (*i.e.* supplementation) of and/or alternatives to individuals' existing methods of communication. Research examining the communication skills of individuals with AS concludes that irrespective of etiology, their speech rarely develops to the extent that it can be used to meet most communication needs and demands (Dan, 2009; Gentile *et al.* 2010; Williams, Peters, & Calculator, 2009). Still, it is very important that we not assume any particular individual with AS cannot learn to use speech as one of many methods of communication. This is particularly true for individuals whose etiologies of AS do not involve deletions. An undetermined number of these individuals can indeed develop functional speech and perhaps even use speech as one of their primary methods of communication. As such, communication programs for individuals with AS should not only target AAC but also speech.

Sometimes concerns are raised about the possibility that providing individuals with AAC will discourage them from speaking – 'why speak if I already have another means of communication?' However, there are no data to suggest AAC

negatively impacts speech in individuals with complex communication needs, including those with AS. To the contrary, any effects on speech tend to be positive (Blischak, Lombardino, & Dyson, 2003; Millar, Light, & Schlosser, 2006).

Most individuals who acquire some functional speech find this method is ineffective and insufficient in certain contexts. As indicated in chapter three, Broad Considerations Driving AAC and Related Instruction for Individuals with Angelman Syndrome by Stephen Calculator, individuals with AS need to be able to communicate with listeners who are and are not familiar with them and their methods of communication. Individuals may find highly familiar communication partners such as family members and one-on-one paraprofessionals or aides understand much of their speech. However, communication breakdowns occur frequently when they use this method to communicate with people who are relatively unfamiliar with them.

It is important to point out that in order to maximize their independence and autonomy, individuals need to be able to communicate effectively with a broad range of people. These include not only family and friends but also acquaintances, paid workers, and unfamiliar partners (Blackstone & Hunt Berg, 2003). Also, while speech may serve an important role, challenges experienced by individuals with AS require their having access to AAC methods that serve other purposes, such as writing, a subject covered by Erin Sheldon in Chapter nine, Comprehensive Literacy Instruction for Students With Angelman Syndrome.

AAC as an Augmentative Method

As noted above, AAC systems are often used to supplement individuals' existing methods of communication. Investigators have found that like individuals whose disabilities are linked to other etiologies, as well as typically developing children and adults, individuals with AS rely on multiple methods of communication (Alvares & Downing, 1998; Calculator, 2013a, b; Calculator, 2014; Martin, Reichle, Dimian, & Chen, 2013; Williams, Peters, & Calculator, 2009). These include a combination of unaided (*e.g.* natural gestures) and aided (*e.g.* communication devices) methods.

To the extent individuals use their existing, or extant, methods of communication

CHAPTER 5

Fostering Communication Skills in Beginning Communicators

Jeff Sigafoos[1,*], Vanessa A. Green[1], Robert Didden[2], Mark F. O'Reilly[3], Giulio E. Lancioni[4], Peter B. Marschik[5,6]

[1] *Victoria University of Wellington, New Zealand*

[2] *Radboud University Nijmegen, Nijmegen, The Netherlands*

[3] *The University of Texas at Austin, Austin, TX, USA*

[4] *University of Bari, Bari, Italy*

[5] *Medical University of Graz, Graz, Austria*

[6] *Karolinska Institute, Stockholm, Sweden*

Abstract: This chapter aims to describe communication assessment and intervention strategies for individuals with Angelman syndrome (AS) who are at the beginning stages of communication development. The chapter begins with a description of beginning communicators. This is followed by an overview of research that has investigated the forms and functions of pre-symbolic behavior (*i.e.*, beginning communication skills) among individuals with AS. Assessment procedures for identifying pre-symbolic forms and functions are outlined. The final section describes intervention approaches for enhancing the effectiveness of pre-symbolic behavior and fostering the transition from pre-symbolic to symbolic communication.

Keywords: Angelman syndrome, Augmentative and alternative communication, Beginning communicators, Enhancement approach, Interpretive approach, Pre-symbolic communication, Replacement approach.

INTRODUCTION

The purpose of this chapter is to describe assessment and intervention approaches that have been applied successfully to foster communication skills in beginning communicators. Many individuals with Angelman syndrome (AS) could be

[*] **Corresponding author Jeff Sigafoos:** Victoria University of Wellington, School of Education, PO Box 17-310 Wellington, New Zealand; Tel: +64 4 463 9772; E-mail: jeff.sigafoos@vuw.ac.nz.

Stephen N. Calculator (Ed)

described as beginning communicators due to the severe nature of their communication impairment. Indeed, one of the defining characteristics of AS is severely limited or absent speech (Angelman, 1965). Because severely limited or absent speech development is characteristic of individuals with AS, many such individuals are candidates for the types of assessment and intervention approaches described in this chapter.

Our description of assessment and intervention approaches for beginning communicators includes consideration of the evidence base underlying these approaches. This consideration is intended to assist parents, teachers, behavior analysts, speech-language pathologists, and other relevant professionals in making evidence-based decisions related to beginning a communication intervention program. As noted by Yell and Rozalski (2013), the Individuals with Disabilities Education Improvement Act (IDEA, 2004) requires the use of empirically validated educational practices for students with disabilities. Empirically validated in this context refers to assessment and intervention procedures that have been shown to be effective in high quality research. Apart from compliance with the law, interventions based on sound research evidence are more likely to be effective (Cook, Tankersley, & Landrum, 2013).

Using empirically validated assessment and intervention strategies could be seen as one way of maximizing the probability of intervention success. Intervention success would seem especially important for beginning communicators and for the beginning stages of communication intervention. An initially positive intervention experience is likely to increase the participant's motivation to cooperate in future intervention efforts. This, in turn, could facilitate the learning of new and more advanced communication skills during subsequent intervention stages, which would in turn greatly enhance the person's social/communication interactions with others. An initially negative experience, in contrast, could seriously dampen the person's motivation to participate in intervention sessions. This, in turn, could negatively impact the learning of new communication skills and thus severely limit the person's overall quality of life.

Ferguson (1994) argued that communication is critically important for enabling meaningful participation. In line with this, Sigafoos, Arthur-Kelly, and Butterfield

(2006) suggested that AAC intervention may enable people with severe/profound disabilities to communicate more effectively with family, friends, teachers, and members of the wider community. For these reasons, it is crucial to ensure that the initial intervention experience is both positive and successful. An important assumption underling this chapter is that positive and successful intervention experiences are more likely when parents and professionals work together to implement assessment and intervention approaches that research has demonstrated to be successful for beginning communicators and for the beginning stages of communication intervention.

In keeping with the AAC overview provided by Stephen Calculator in Chapter 4, the main focus of this chapter is to describe approaches related to the use of one or more augmentative and/or alternative (AAC) modes of communication. A range of non-speech methods of communication is considered under the umbrella of AAC intervention. Gestures/manual signs, picture-exchange communication, and/or the use of electronic speech-generating devices are potential options, but several other AAC modes are often indicated for the beginning communicator. One additional option that is highly relevant for beginning communicators aims to make use of and enhance the person's existing pre-symbolic behaviors.

To make effective use of existing pre-symbolic behavior it is necessary to first identify the existing pre-symbolic communicative forms and functions that are already present in the repertoire of the beginning communicator. Assessment approaches have been developed that appear effective in identifying existing pre-symbolic communication behavior. This assessment information is then used for planning the beginning stages of communication intervention. At the beginning stages of communication intervention, efforts have been successfully directed at (a) enhancing the communicative effectiveness of the person's existing pre-symbolic behaviors, and/or (b) facilitating the person's transition from pre-symbolic communication to the use of symbolic forms of communication, such as unaided AAC (*i.e.*, manual signs/gestures) or aided AAC (*e.g.*, selecting line drawings or photographs from the display of a speech-generating device).

To set the stage for beginning an AAC intervention, this chapter is structured into several sections. The next section aims to define and describe the characteristics

CHAPTER 6

Fostering Effective Communication in Individuals With Angelman Syndrome Who Rely on Symbolic Methods of Communication

Stephen N. Calculator*

Department of Communication Sciences and Disorders, University of New Hampshire, Durham, NH, USA

Abstract: This chapter discusses principles as well as assessment and intervention strategies designed to foster effective communication skills in individuals with Angelman Syndrome who rely on both pre-symbolic and symbolic methods of communication. Both unaided and aided methods of augmentative and alternative communication are considered. By virtue of the nature of their disabilities, coupled with recent research findings, all individuals with AS are not only deemed candidates for AAC but also potential users of high-tech AAC devices. A review of best practices related to providing AAC services to individuals with AS is presented along with information pertaining to parents' corresponding priorities.

Keywords: AAC, Alternative communication, Angelman Syndrome, Assessment, Augmentative, Best practices, Disability, Education, Generalization, Genetics, Handicap, Intellectual, Intervention, Outcomes, Parents, Sign language.

INTRODUCTION

As discussed by Stephen Calculator in chapter seven, in conjunction with Enhanced Natural Gestures, many individuals with AS rely on relatively basic, highly concrete methods of communication that are sometimes referred to as pre-symbolic. Conversely others use systems that include various symbols, unaided as well as aided, and are thus characterized as symbolic communicators. This chapter will focus on the latter. It is important to point out however that there is no clear

* **Corresponding author Stephen N. Calculator:** University of New Hampshire, Department of Communication Sciences and Disorders, Hewitt Hall, Durham, NH 03824, USA; Tel: (603) 862-3836; Fax: (603) 862-4511; E-mail: Stephen.calculator@unh.edu.

line that separates individuals who use pre-symbolic methods from those relying on symbolic means of communication. In fact, as described later in this chapter, even those individuals who access high-tech AAC devices often rely concurrently on presymbolic methods such as natural gestures, nonspeech vocalizations, and physical manipulations of people and objects.

Readers are encouraged to review the contents of chapter four, which provides an overview of AAC, before proceeding to this chapter, as it introduces several key terms and principles that are then explored further here. This chapter begins with a discussion of the principle of zero exclusion as it applies to individuals' eligibility for AAC services. This principle is then applied to unaided methods of communication such as natural gestures, sign language, and Enhanced Natural Gestures. Relative benefits and drawbacks of these systems for individuals with AS are discussed and options to address the latter are presented. Next, the principle of multimodal communication is introduced, with readers encouraged to consider output as well as input modes.

The chapter then shifts focus to aided communication. An argument is made for assuring all individuals with AS access to high-tech devices, once again applying a standard of zero exclusion. Readers are encouraged to adopt the principle of the 'least dangerous assumption' to avoid scenarios in which individuals are denied access to such aids based on assessed cognitive, linguistic, and other capabilities, only to find at a future date that such systems would likely have been effective if introduced earlier.

The process of generating useful outcomes of assessment is discussed in relation to the need for multidisciplinary supports for AAC. A call is made for assessment and intervention procedures that are ecologically valid, with several examples given. Professionals are encouraged to think beyond the boundaries of their respective disciplines, sharing a common set of goals and visions. Practices that support such efforts are discussed.

This leads to a review of functional outcomes of AAC. Research examining factors associated with individuals' acceptance and rejection of AAC devices is reviewed, and possible best processes are articulated in a general sense as well as

in relation to the four components of AAC systems; symbols, techniques, devices and other methods of communication, and strategies.

Zero Exclusion Principle

In light of the fact that all individuals with AS exhibit severe communication challenges, with few able to use speech as a primary method of communication, all should be considered candidates for AAC systems. There are no cognitive, motor, language, sensory or other pre-requisite skills individuals with severe disabilities must possess in order to be considered for an AAC system (Kangas & Lloyd, 1988; Romski & Sevcik, 1988). Individuals should not be considered 'too something' or 'not ready for' to rule them out as candidates for AAC (Beukelman & Mirenda, 2013). Types of AAC devices and systems that are applicable for individuals certainly vary depending on factors such as individuals' capabilities and needs; characteristics of communication partners; and settings in which interactions occur. However the question is not whether or not an individual with AS would benefit from AAC but instead what systems would be most beneficial and how might they be introduced to maximize communicative success.

Individuals with AS have been found to rely concurrently on a variety of methods of communication in meeting their daily communication needs (Calculator, 2013a, b; Calculator, 2014; Hyppa Martin, Reichle, Dimian, & Chen, 2013). These include unaided as well as aided systems. They also use a combination of pre-symbolic methods, such as natural gestures, physical manipulations of people and objects, and nonspeech vocalizations, as well as symbolic means such as photographs, picture communication symbols and printed words that are affixed to speech generating devices.

Unaided Communication

A sizeable proportion of individuals, especially those who are deletion negative, use sign language as one of their many means of communication (Calculator, 2013a). This may be problematic in light of findings that a large percentage of signs, even when produced accurately, cannot be understood by others (Lloyd & Karlan, 1984; Rotholz, Berkowicz, & Burberry, 1989). Signing systems vary from country to country, thus there is British Sign Language, American Sign Language,

CHAPTER 7

Enhanced Natural Gestures: A Self-Administered Program for Teaching Communicative Behaviors to Individuals with Angelman Syndrome

Stephen N. Calculator*

Department of Communication Sciences and Disorders, University of New Hampshire, Durham, NH, USA

Abstract: The ultimate goal of this chapter is to provide parents, speech-language pathologists, educators, personal care assistants, and others with a sufficient orientation to Enhanced Natural Gestures (ENGs) to enable them to administer the program on their own. ENGs are intended to serve as one component of a broad augmentative and alternative communication (AAC) system that includes multiple modes of communication. ENGs consist of intentional behaviors that individuals are already using or can be easily taught given their existing cognitive, language, and motor skills. Unlike other gestural systems, including sign languages, ENGs are easily acquired by individuals with AS and readily understood by a broad range of communication partners. ENGs often differ from one individual to the next. Readers are acquainted with the two primary methods for teaching ENGs: Mand-Model with Time Delay (MMT) and Molding-Shaping. These techniques take advantage of naturally arising opportunities for communication that occur at school, home, and other settings.

Keywords: AAC, Alternative, Angelman Syndrome, Augmentative, Communication, Candidacy, Disability, Education, Enhanced, Genetics, Gestures, Handicap, Intellectual, Intervention, Natural, Severe, Sign, Speech, System.

INTRODUCTION

This chapter provides a step-by-step description of how to teach Enhanced Natural Gestures (ENGs) to individuals with Angelman Syndrome (AS). Like individuals without disabilities, individuals with AS have been found to use

* **Corresponding author Stephen N. Calculator:** University of New Hampshire, Department of Communication Sciences and Disorders, Hewitt Hall, Durham, NH 03824, USA; Tel: (603) 862-3836; Fax: (603) 862-4511; E-mail: Stephen.calculator@unh.edu.

Stephen N. Calculator (Ed)

various modes of communication concurrently (Calculator 2013a, b). These usually include both unaided (*e.g.* natural gestures, sign language, non-speech vocalizations, and speech) as well as aided (*e.g.* physical manipulations of people and objects; Pragmatic Organization Dynamic Displays, or, PODDs Porter, 2007; communication boards; and low and high tech electronic devices) methods. ENGs are rarely intended to replace other methods by which individuals are already communicating successfully. One notable exception involves cases in which ENGs are taught as alternatives to challenging behaviors (*e.g.* hitting, scratching, and pulling hair). ENGs are more likely introduced as augmentative, or, supplementary methods of communication.

SOCIAL NETWORKS

Chapter three discussed the concept of social networks (Blackstone & Hunt Berg, 2003a, b; Calculator & Jorgensen, 1992) as an important construct to attend to in designing communication and related programs for individuals with AS. Parents and practitioners were encouraged to teach individuals communication skills that foster their abilities to interact and develop relationships with a broad range of communication partners. These include individuals who are paid to be in their lives, such as teachers, therapists, and employers, as well as unpaid communication partners such as family and friends. Some communication partners may be familiar with individuals with AS as well as their methods of communication. For example a child's aide, someone familiar with Molly and her methods of communication, may know that when Molly strikes her stomach she is indicating she has lost interest in an activity and wants to move on to something different. Molly may find this communicative behavior is usually successful with her aide. Conversely, if her aide is sick one day and is replaced by another staff who is unfamiliar with Molly and her methods of communication, that person may be unable to respond successfully to this same behavior.

CHALLENGES OF SIGNING

Individuals with AS, especially those without deletions, may use sign language as one of their primary methods of communication (Calculator, 2013a). If they use signs from American Sign Language (ASL) accurately, they are likely to be able

to communicate with both familiar and unfamiliar communication partners who have background in ASL. If they are to be successful communicating with partners unfamiliar with sign language, their communicative success depends on their abilities to supplement ASL with other unaided (*e.g.* natural gestures) and aided (*e.g.* communication devices) methods of communication.

Taking this one step further, individuals with AS who sign often lack the motor skills necessary to produce signs accurately (Calculator, 2002). Instructors may compensate for this difficulty by introducing modified signs that bear varying levels of similarity to the original versions. Unfortunately, their signs may now no longer be intelligible to unfamiliar persons irrespective of whether or not they have formal signing background. Only persons familiar with these individuals and their modified signs may be able to understand and respond appropriately to their communicative attempts. Social networks of such individuals may thus be very limited.

Previous investigators have documented the fact that even when they have access to sophisticated electronic communication devices, individuals with AS often prefer to use simpler, nonsymbolic methods of communication such as natural gestures and non-speech vocalizations. Calculator (2013b) reported parents of individuals with AS cited such methods as important components of their children's AAC systems for several reasons, the primary one being their ease of use. Signing was rarely cited as a component of children's most effective methods of communication, regardless of their having accepted or rejected AAC devices, though it was more prominent in the latter situation (8% v. 20%, respectively).

Given the language, cognitive, and motor limitations characteristic of individuals with AS, it is not surprising that signing presents significant challenges to them. As noted above, compensations often take the form of modified signs. Given the limitations imposed by signs (*e.g.* restricted social networks), modified or not, there appeared to be a need for an alternative system. The latter would appear particularly viable if it built upon children's natural preferences for and propensities to use natural gestures. The Enhanced Natural Gestures program was developed with this in mind.

CHAPTER 8

Application of Principles of Applied Behavior Analysis in Addressing Challenging Behaviors of Individuals with Angelman Syndrome

Jane Summers*

Centre for Addiction and Mental Health, Toronto, Ontario, Canada

Abstract: This chapter provides an overview of challenging behavior in children and adults with Angelman syndrome and outlines possible reasons regarding how and why it may develop. Approaches to address challenging behavior that are based on the principles of applied behavior analysis are presented, along with ways to replace challenging behavior with socially acceptable skills. Case examples are used to illustrate assessment and intervention procedures, with a focus on prevention of challenging behavior and functional communication training.

Keywords: AAC, Aggression, Alternative communication, Angelman Syndrome, Antecedents, Applied behavior analysis, Augmentative and alternative communication, Behavior, Challenging behavior, Consequences, Disability, Functional assessment, Functional communication training, Genetics, Skill deficits.

INTRODUCTION

The contents of this chapter complements that provided by Jeff Sigafoos and colleagues in chapter 5, Fostering Communication Skills in Beginning Communicators, which considers uses of applied behavior analysis procedures in teaching replacement behaviors to individuals with particularly complex communication challenges. The current chapter will begin by providing information about the learning and skill profile of children[1] with Angelman

* **Corresponding author Jane Summers:** Ph.D., Centre for Addiction and Mental Health, Underserved Populations Program, 80 Workman Way, Room 5232, Toronto, ON, Canada M6J 1H4; E-mail: jane.summers@camh.ca.

[1] Although references are made to children with Angelman syndrome throughout the chapter, most of the points that are raised also apply to adults with AS.

Stephen N. Calculator (Ed)

syndrome, a subject also discussed by Erin Sheldon in chapter 2, Learning Characteristics of Students With Angelman Syndrome and Related Instructional Strategies. This will be followed by an overview of challenging behavior in children with AS and a discussion of possible reasons regarding how and why it may develop. The next section will outline approaches to address challenging behavior that are based on the principles of applied behavior analysis, followed by a section on replacing challenging behavior with new skills to increase children's personal and social competence. Throughout the chapter, case examples will be used to illustrate key points.

Behaviorally-trained professionals recognize the importance that other specialists play in the lives of children with AS, including the neurologist, psychologist and psychiatrist, speech-language pathologist, occupational therapist, physiotherapist, and special education teacher, among others. Their input is respected and the need to coordinate efforts to address challenging behavior with the work they do is acknowledged.

LEARNING AND SKILL PROFILE OF CHILDREN WITH AS

Certain characteristics of children with AS may affect their ability to learn new skills and contribute to the development of challenging behavior. Many of these characteristics are found commonly among the broader population of children with severe intellectual disabilities, such as the tendency to become frustrated in learning situations, or being difficult to motivate. Some characteristics, however, are more specific to AS. One of the universal features of children with AS is a movement and balance disorder (Williams *et al.*, 2006). Many children have jerky or unsteady movements. They also have difficulty imitating people's actions (Summers & Szatmari, 2009). These motor impairments can make it difficult for them to perform actions that require motor planning and fine and gross motor coordination such as lifting their arms up over their head to put on a shirt or bringing a spoon to their mouth. Since most responses involve some type of motor component, this is a particularly important area to target for remediation.

Another common characteristic of many children with AS is hypermotoric behavior (Williams *et al.*, 2006). They seem to be constantly "on the go"

(Williams, 2010) and show signs of restlessness and distractibility, which can impact their ability to concentrate in learning situations (Tan *et al.*, 2011).

The majority of children with AS have a seizure disorder and are taking anticonvulsant medication to control their seizures (Clayton-Smith, 2010). The degree of seizure control can vary from time to time and break through seizures can occur. Some children can experience absence seizures many times during the day. This seizure activity can make them seem less responsive and recurrent, prolonged seizures may affect their ability to learn (Thibert *et al.*, 2013). Anticonvulsant medication may cause unpleasant side effects such as fatigue, lethargy and irritability (Thibert *et al.*, 2009) and can add to children's cognitive impairment. However such treatment is essential from the standpoint of controlling their seizure activity.

Many children with AS have sleep disturbances and a decreased need for sleep (Pelc, Cheron, Boyd, & Dan, 2008). This often results in them, as well as family members, being tired (Goldman, *et al.*, 2012) and can have an impact on their learning performance and behavior (Bruni *et al.* 2004). Fatigue may be accompanied by physical signs, such as dark circles under children's eyes, as well as behavioral changes like being less cooperative and attentive or showing signs of irritability by protesting or "complaining". Like anyone else, children with AS may not learn as effectively when they are excessively tired, are having more seizures, or are not feeling well.

Impairments in expressive language, and to a somewhat lesser degree, receptive language, are a universal feature of children with AS (Williams *et al.*, 2006) and have a profound impact on their day-to-day functioning. For the vast majority of children with AS, speech alone is not a viable mode of communication (Andersen, Rasmussen & Stromme, 2001; Didden *et al.*, 2009). Augmentative and alternative communication (AAC) approaches can be helpful for many children who do not develop spoken language skills or as a means to enhance the expressive abilities of those who do have some minimal speech skills (Calculator, 2002, 2013a,b; Summers *et al.*, 1995). Different examples of AAC approaches include using manual signs, tangible symbols (real or miniature objects), representational symbols such as photographs and picture communication symbols, and high and

CHAPTER 9

Comprehensive Literacy Instruction for Students with Angelman Syndrome

Erin S. Sheldon*, Caroline Ramsey Musselwhite

The Angelman Literacy Project Kingston, Ontario, Canada

CCC-SLP Assistive Technology and Literacy Consultant Phoenix, Arizona

Abstract: Most students with Angelman syndrome (AS) are at an emergent level of language and literacy development. Students with AS require the same comprehensive literacy instruction as their peers with typical development, but they need specific adaptations, supports, and opportunities in order to access that instruction. Comprehensive emergent literacy instruction engages students with AS in using symbolic language for authentic purposes: to comprehend and respond to the stories of others and to express their own ideas and individuality. This comprehensive instruction develops observable skills and understandings in the areas of vocabulary, reading comprehension, reading fluency, phonemic and phonetic awareness, and writing.

Keywords: AAC, Accommodations, Alternative assessment, Angelman syndrome, Complex communication needs, Comprehensive, Education, Emergent literacy, Fluency, Generative writing, Language, Literacy, Modifications, Phonemic awareness, Phonics, Portfolio assessment, Reading comprehension, Shared reading, Shared writing, Symbolic, Vocabulary, Writing.

INTRODUCTION

In chapter two, we described how the nature of the disabilities associated with Angelman Syndrome influences learning in the classroom. In this chapter, we explore how the nature of these disabilities specifically impacts literacy instruction. As this chapter explains, all students can grow in their emergent literacy understandings when provided access to personally meaningful opportunities to engage with print, language, and tools for writing and

* **Corresponding author Erin S. Sheldon:** The Angelman Literacy Project, 37 Inverness Crescent, Kingston, ON, K7M 6P2, Canada; Tel: (613) 328-7632; E-mail: sheldon.erin@gmail.com.

Stephen N. Calculator (Ed)

communicating.

My daughter Maggie (see Fig. **9.1** and **9.2**) is 11 years old. She has bright copper curls, a wide smile, and bubblegum pink Doc Martens boots that she insists on wearing every day. She has a large deletion on her 15th chromosome, resulting in Angelman Syndrome (AS). Maggie attends our neighborhood school in a regular 6th grade class. When she was little, her individualized education plan emphasized simple functional life-skills goals while encouraging her to participate in classroom activities. As she got older, her school inclusion required more problem solving about how Maggie could participate meaningfully in the increasingly complex general curriculum.

Fig. (9.1). Maggie age 11, Angelman Syndrome, Deletion positive.

Fig. (9.2). Maggie with her Girl Guide unit.

This problem solving led me to graduate school in the field of Education, where I studied literacy acquisition for students with the most complex disabilities. Maggie was in 2nd grade when I began researching how to better include and educate her. At this age, Maggie selected from a binder of photographs to request preferred activities in each subject, such as the parachute in gym or magnetic blocks in math. She had several applications on her iPad™ for augmentative and alternative communication (AAC) but she didn't use them. She used her iPad to watch home movies and popular music videos, particularly her beloved Justin Bieber. Maggie's AAC assessment described her as pre-communicative. She demonstrated minimal interest in books and would often rip pages, but would browse books that had touch-and-feel elements or large photographs of Justin Bieber. She was reluctant to grasp markers or pencils and made only the barest marks on paper. Maggie had a stamp for her name but often resisted the hand-over-hand assistance she required to label her work.

When Maggie was in 4th grade, we shifted her individualized education plan to

CHAPTER 10

The Role of AAC In Fostering Inclusion of Adults with Angelman Syndrome in Post-School, Home and Community Settings

Teresa Iacono*, Hilary Johnson

La Trobe University, Australia

Abstract: The communication of adults with severe intellectual disability is difficult to characterize. The literature suggests their skills fall into stages of pre-symbolic to symbolic or even basic linguistic skills. Recent research, however, suggests a more complex picture of combining pre-symbolic and symbolic forms used within strategies that vary according to social relationships. In this chapter, the communication characteristics of people with severe intellectual disabilities are explored according to the research literature, with a focus on social interaction processes that value the person's extant skills. With this literature as a background, the use of AAC to enhance inclusion of people with severe intellectual disability, and in particular adults with Angelman Syndrome, across settings, such as community, home and day service, and work will be explored.

Keywords: AAC, Adults, Alternative communication, Angelman Syndrome, Assessment, Augmentative, Day services, Disability, Intellectual disability, Intervention, Post-school, Social inclusion, Social relationships, Supported accommodation, Transition.

INTRODUCTION

As individuals with Angelman Syndrome (AS) transition from school to community settings of day services or work, they face challenges that extend well beyond those experienced by most people in moving into an adult world. Although children and adults with AS show a variable cognitive profile, related to some extent to their underlying genetic characteristics (Williams *et al.*, 2006),

* **Corresponding author Teresa Iacono:** La Trobe Rural Health School, PO Box 199, Bendigo, VIC 5440, Australia; Tel: (613) 5444-5448-9110; Fax: (613) 5444-9199; E-mail: t.iacono@latrobe.edu.Au.

Stephen N. Calculator (Ed)

severe to profound intellectual disability and minimal functional speech have been identified as frequent co-existing disabilities, resulting in communication that is commensurate with beginning communicators (Calculator & Diaz-Caneja Sela, 2015). Given this profile, strategies to support the school to community transition and functioning in contexts that contrast sharply to school have been gleaned from a literature base that is broadly focused on severe intellectual disability rather than more narrowly on the characteristics and experiences of young adults with AS. In this chapter, the challenges faced by adults with AS as they transition from school to post-school are informed by literature of relevance to adults with severe intellectual disability. These challenges provide the context in which to consider the role of social relationships in providing opportunities for communication through the use of AAC and other strategies to facilitate social inclusion across home and day activity settings.

Communication Characteristics

Scoping the literature on AS, communication and AAC reveals varied communication profiles that span pre-intentional to emerging symbolic stages associated with young children in their first 18 months of life (Calculator & Diaz-Caneja Sela, 2015). As noted by Calculator (Chapter 3), such developmental comparisons lack meaning as individuals move from early childhood, to adolescence and then adulthood, in light of the complex interplay of life experiences and a varied underlying skills base. Hence, individuals who may lack basic symbolic ability, but readily engage in social interactions will learn to use any modalities or strategies within their abilities to engage with their physical and social environments.

Young Children

Research with older individuals with AS is lacking; there is a much greater focus on profiling skills of young children. This research demonstrates variability in language expression and production, but with few children with AS whose level of intellectual disability is severe to profound demonstrating the ability to combine symbols with sufficient flexibility and variation to indicate linguistic ability. There is also evidence of particular problems with gestural communication, although the work of Calculator (2002) on enhanced natural gestures has

demonstrated the benefits of focusing on extending gestures within a child's repertoire to an expanded range of communicative functions.

Adults

Data about adults with AS has tended to be included in large scale studies with participants ranging in age from young children to older adults. Didden, Korzilius, Duker, and Curfs (2004), for example, assessed the expressive communication of 109 participants with AS, aged from 2 to 44 years. Their findings indicated greater strength in using communicative requests than comments, and co-morbidities, such as epilepsy, being associated with poorer communication. Unfortunately, this and other studies including adults with AS have not differentiated results according to age groups or degree of intellectual disability (Calculator, 2013; Didden *et al.*, 2009). Nonetheless, the evidence provided reflects profiles of skills seen in adults with severe intellectual disability more generally (L. McLean, Brady, McLean, & Behrens, 1999; Snell *et al.*, 2010).

The communication of adults with severe intellectual disability is difficult to characterize in light of its variability. Research by L. McLean *et al.* (1999) indicated such wide variability across 84 children and adults with severe intellectual disability. Adults in their study relied largely on non-symbolic intentional communicative behaviors, including vocalizations and gestures that were produced on or close to a referent object or communication partner (contact) or at a distance from a referent object or communication partner (distal), with some also producing single or a combination of conventional symbols (words, signs, graphic symbols). The functions of their communication were largely requests (imperatives), with few comments (declaratives) evident. Participants who used largely contact gestures produced few communicative acts overall, while those using distal gestures communicated as frequently as those using single or combined symbols. Similarly, in a recent review of communication intervention research of people with severe intellectual disability, Snell *et al.* (2010) noted that pre-intervention skills across 116 studies were characterized mostly as pre-linguistic or emerging.

EPILOGUE

Stephen N. Calculator*

Department of Communication Sciences and Disorders, University of New Hampshire, Durham, NH, USA

This book began with prologues writtten by three parents of two children and one adult with Angelman Syndrome. All three of these parents have made substantive contributions to the Angelman community in conjunction with more personal efforts advocating for quality services for their children. One primary purpose of these prologues was to enable parents to share their experiences, positive as well as negative, with other parents as well as professionals. Unless both groups are 'on the same page' so to speak, working for a common set of goals and maintaining a single unifying vision, prospects for providing individuals with AS the supports they need to not only achieve but excel in education and life may be guarded at best. Subsequent chapters were intended to expound on issues to consider and actions that might be taken to actualize these parents' and others' aspirations for their chidren. Many of the practices that were deliniated are not bound by individuals' intellectual and other restricitons but can instead be applied universally not only to individuals with severe disabbilities but typical children and adults as well. Let's take a moment to revisit some themes one or more parents articulated in their prologues.

Coping With the Diagnosis

All three parents provided accounts of how they received and then reacted to the news that their children had Angelman Syndrome. What began as grief and sometimes confusion eventually transcended into visions and development of action plans their children would require. Parents found great comfort interacting with professionals who looked beyond the diagnosis of AS and instead viewed their children as unique individuals whom, with the appropriate supports, could achieve great things at school and elsewhere. Much of the literature on AS can be both daunting and discouraging as a menu of disabilities and devastating implications are recounted. These parents were able to see beyond their children's disabilities, focusing instead on their special abilities. Amy Girouard expressed this nicely in commenting, "For us, it was an important lesson in looking at Ally as an individual and not the diagnosis."

* **Coresponding author Stephen N. Calculator:** University of New Hampshire, Department of Communication Sciences and Disorders, Hewitt Hall, Durham, NH 03824, USA; Tel: (603) 862-3836; Fax: (603) 862-4511; E-mail: Stephen.calculator@unh.edu.

Garnering Resources

Parents' and their childrens' successes at school and elsewhere were contingent on locating the resources children needed. This included access to professionals who believed in children's potentials and recognized the roles they might play in fostering positive outcomes. Susan Ravellette speaks of the point at which her families' lives, "became inundated with various therapies, agencies, and paperwork." Profesionals must reognize the impact their involvement may have not only on indivdiuals with AS but also their families. What are the pushes and pulls on family life and how much can we expet families to adjust their lives to accommodate professionals' expectations?

Maintaining High Expectations

These parents maintained high expectations of their childrren, despite their diagnosis of AS. They recognized their children were special and yet this did not absolve them from being held responsibile for serving important roles in their families' daily lives, including carrying out daily chores. This carried over to school as well, where parents acknowledged their children faced special challenges but did not accept this precluding them from being challenged acadmically and otherwise.

Planning for the Future

Perhaps Eileen Braun articulated this best in her comment, "I always had in mind how will what we are doing here, today, get us to the day the bus stops coming?" This certainly has numerous implications for program design and implementation. It is imperative that education foccus on skills individuals will require to live happily and independently as possible post-school. This is not restricted to activities of daily living such as eating indepently, using public transportation, using the bathroom, and acquiring vocational sklls, but instead includes skills such as reading, writing, and effecive uses of high-tech AAC devices and other methods of communication. Inclusive education is not coneptualized soley as being in the same place as typical peers but as importantly a set of contexts in which children's individualized educational needs can be addressed. School and the eventual transition to community life, and work, requires acquisition of skills with functional, long-term outcomes.

Maintaining a Sense of Normalcy

Parents talked about the impact the initial diagnosis of AS, and subsequent challenges faced by all family members as they found their lives suddenly revolving in large part around one child's needs. Professionals are encouraged to recognize the impact of AS on families and offer families supports they need to function as a well-tuned family unit. Families cannot be

defined by AS. Instead AS co-exists in an environment that is sensitive to each familiy member's needs and desires. Life must not revolve exclusively around the diagnosis of AS. Instead families should seek out opportunities to engage in activities necessary to sustain and grow the quality of their own lives. As noted by Amy Girouard, this includes taking time for intimacy with one's spouse or partner.

The title of this book, "Angelman Syndrome: Communication, Educational, and Related Considerations" suggests a unique set of knowledge and skills professionals must posess in order to meet the needs of individuals with AS and their families. In her opening parent prologue, Eileen Braun communicated the urgency of doing so, commenting, “As educators, you have this year, maybe the next year—but the families have the next 40, 50 or 60 or more years with their child with Angelman syndrome. Have a sense of urgency. We can’t afford another week, two weeks, a month, a semester, or next year to get something in place for our kids. There are no ‘mulligans’ or do-overs—now is the time.” It is our hope that this book has accomplished a primary aim of better equipping its readers to take on this challenge and to do so in a timely manner.

SUBJECT INDEX

P

R

S

T

U

V