IS WILLIAMS SYNDROME THE 'CONVERSE' OF AUTISM SPECTRUM DISORDER?

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Abstract: Williams Syndrome (WMS) and Autism Spectrum Disorder (ASD) are neuro-developmental disorders associated with distinct social phenotypes. Infant and young children associated with these two disorders provided evidence of developmental delay. This paper attempts on analyzing the extent of convergence and divergence of the behaviours in WMS and the symptoms of ASD. Features of diagnosis and characteristics, as well as developmental achievements in these two disorders will also be addressed further. Moreover, in depth discussion and comparison on a number of aspects including; intellectual ability, visual-spatial skills, language and communication, social interaction and emotion detection, in regards to both disorders will be dealt in detail throughout the rest of this paper.

INTRODUCTION

Williams Syndrome

Children with Supra-Valvular Aortic Stenosis (SVAS) were referred to as Williams Syndrome (WMS), coined by Williams and colleague in the early 1960s (William & Campbell, 1960). These children were characterized with several physical appearances, which consist of medial flare of the eyebrows, lacy stellate (star-burst) pattern of the irises, flat nasal bridge with a bulbous tip and ante-verted nares. Their facial look is often described by the founder as elfin or 'pixielike' and symptoms of mental retardation were also reported (William & Campbell, 1960). Interestingly, similar characteristics were found in a study conducted by Beuren and colleague (1962). In their study, they identified another set of feature in which the children have hypoplastic teeth despite full lower lip with a wide smile, and small chin (Beuren, Apitz & Harmjanz, 1962).

They are also characterized as being very friendly and active. For example, von Arnim and Engel (1964) described individuals with WMS as showing a very good rapport with anyone they meet and easily involved in the conversation with their communication partner. They were able to establish interpersonal contacts within a few minutes time. Those being identified with WMS seems like very curious of getting to know each without any worries even with strangers. Kaplan et al. (2001) had listed four characteristics that have elicited the most interest which are the individual's social relationships, activity level, anxiety level, and sleep problems. The most consistent outcomes have been reported is that the children with WMS are highly interested in social interactions and very empathic.

In some cases, other personality characteristics appeared to be such as hyperactivity, impulsivity and low frustration tolerance (Tomc, Williamson & Pauli, 1990). Therefore, it may cause problems with social interactions. Along these lines of reasoning, researchers have found that individuals with WMS have difficulty in understanding other's beliefs or points of view, which may contribute to some of their social difficulties. Individuals with WMS are also reported to present low IQ levels (Mervis, Morris, Bertrand & Robinson (1999). Despite the case, their cognitive functioning including vocabulary knowledge, face processing, and auditory rote

memory remains intact (Bellugi et al., 1992; Mervis et al., 2000). Interestingly, when compared with other atypical groups (eg. Down syndrome or non-specific mental retardation) that matches their mental age, subjects with WMS were reported to gain significantly higher scores on cognitive abilities (Wang, 1996).

WMS is thought to be due to genetic mutation. Studies revealed that the mutation occurs at the long arm of Chromosome 7. Elastin gene, which is located at the aforementioned chromosome, is micro-deleted in the case of WMS. This deletion often results in severe physical abnormalities including; cardiovascular complications, elevated blood calcium levels, sensitive hearing and high blood pressure, failure to thrive in infancy, abnormal sensitivity to certain classes of sounds (hyperacusis) (Ewart et al.,1993), and moderate to severe learning difficulties (Plissart & Fryns, 1999). In a study conducted by Paul et al. (2000) an estimate of 1 in 20,000 live births suffered WMS and nearly all cases resulted from de novo deletion events in the genetic material.

Autism Spectrum Disorder

Above paragraphs had described briefly on WS and its characteristic. Accordingly, this paper will explain some aspect to be known about ASD before further discussion. Autism is relatively diverse, and to define it properly is rather subjective. Children with ASD are described as without any obvious physical signs and biological markers (Frith, 2003). However, despite the lack of the previously mentioned characteristics, it is very similar to WMS in terms of mental retardation or learning disability. The developmental approach of cognitive theory referred to as the *mind-blindness hypothesis* explains the underlying features of autism. This theory proposes that, children with ASD have missing their intuitive ability to understand other people's thoughts and mental state (Baron-Cohen, 1995). In fact, previous study assumed that ASD as resemblance to an adulthood mental disorders named schizophrenia (Wing & Gould, 1979). However, since a question arises about the children who are diagnosed with schizophrenia before puberty, therefore it remains debated.

Despite numerous studies on ASD, the aetiology remains unknown. However, genetic and environmental factors are thought to play important roles in the development of ASD. Environmental factors including complication during pregnancy and birth often contributes to fetal abnormalities that present in children with ASD (Frith, 2003). Children suffered with ASD frequently presented with brain damage in comparison to normal individuals. Viral infection and immunization disorders were also thought to cause ASD however these factors are trivial. In terms of the genetic factor, Rutter and Folstein (1977) reported that approximately 90 percent of concordance was found in identical twins for milder forms of autism. In addition, Rutter (2000) reported that 3 to 6 percent the risk of second child being affected in family that has more than one child with ASD.

Originally referred by Kanner (1943) and Asperger (1944), ASD relates to a fundamental biological disturbance that is present from birth. Both authorities found some fascinating features of these children that seemed they have disturbance to establish normal relationships with the peers. A child with ASD is more interested in the world of objects rather than peers and people around them. Indeed, autistic loneliness, desire for sameness and islets of ability serve as a reference point for features of classic autism (Kanners , 1943). Unlike other typical developing children, Abbe Pierre-Josseph noticed that children with ASD are lack of imaginative play. This deviant characteristic was realized since 1800 towards a boy called Victor (Lane, 1976). All the findings come into logical conclusion that children with ASD appeared to be difficult to participate normally with other people and their surroundings thus, lead to disturbance in social integration.

Children with ASD were initially thought to be deaf. This premise was brought up given the lack of responsiveness to other people's voice. However, it was realized that these children were actually reacted unusually towards certain sound (Rapin & Allen, 1987). Children suffered with ASD are monotonously repetitious in all their performance just like as it was when they utter verbally. Besides that, communication impairment such as delayed speech production was also observed in ASD children (Bauman, 1999). While children with hearing impairment communicate through signs, gestures and facial expressions, children with

ASD tended not to do so. Nevertheless, despite their handicap, these children exhibited unexpected outstanding achievements. These children have an excellent memory and often skilled in music and drawings (Happe 1999).

Using standard diagnostic handbooks, Chakrabarti and Fombonne (2001) showed a huge increase of prevalence approximately 60 per 10 000 of the population with ASD. The increasing prevalence is probably due to greater awareness of autism that meant for more cases looking forward to be diagnosed. Equally important, the increment of the prevalence accordingly reflected by varies definition of autism itself across professional in different countries. Recently, the diagnostic criterion of ASD is described in Diagnostic and Statistical Manual (DSM) (American Psychiatric Association, 2000). In the same way with classical features of autism, it was decided by American Psychiatric Association (APA) (2000), children must at least meet with two out three criteria, which include qualitative impairment in reciprocal social interaction, qualitative impairment in verbal and non-verbal communication relative to developmental level. Also, they must be a markedly restricted repertoire of activities and interests appropriate to developmental level. These criteria have been collectively accepted worldwide for ASD diagnosis and to the benefit of research purposes and clinical practice (Hill & Frith 2003). The following paragraphs will discuss issues that concern with converging aspects in both WMS and ASD.

Overlapping behavioral symptoms in Williams Syndrome and Autism Spectrum Disorder

There are numerous studies that indicate the similarities of WMS and ASD. Symptomology patterns for instance, especially unusual perception and sensitivity towards sound and noise, were found to be overlapped in children diagnosed with ASD and WMS (Levithin et al., 2005). The similarity in terms of genome analysis also has been discussed in years back (Berg et al., 2007). Another study conducted by Lincoln et al. (2002) showed patients with WMS exhibit inability to rapidly shift attention, a characteristic similar to adults with autism and cerebellar lesion patients. In this study, three main tasks were conducted mainly on visual and auditory focus, as well as shifting attention. However, due to individual differences in terms of their cerebellar vermis size, a much longer time was needed to allow them to shift their attention to match the performance present by individuals with ASD. Despite interesting findings, several limitations were found including small number of patients combined with issues concerning age groups. Further study with more parameters could be conducted to get much concrete results.

Additionally, mental retardation or learning disability is found to be frequently associated with both ASD and WMS. Chakrabarti and Fombone (2001) reported that 25% to 40% of patients with ASD have an IQ under 70. Other report showed approximately 25% of children with WMS to have learning disability while the remaining displays unlimited IQs and adaptive behavior quotient consistent with mental retardation (Mervis and Klein-Tasman 2000). However, the exact average cut of point of IQ levels in both disorders varies upon time as they might present in different degree of cognitive ability in each of the disorder. In fact, some of the evidence unlikely would affect the percentage obtained when some of the assessment ware not conducted using the standardized test.

Furthermore, unique ability usually called as savant abilities were known to occur in both disorders. For example, children with ASD displayed extra-ordinary skills in areas of music, arts, and mathematics. As mentioned by Frith and Hill (2003), at least 10% of the autistic population to be present with these set of skills. Similarly individuals with WMS exhibited cognitive strength in music and arts. It is thought that certain areas of the brain are ‘preserved’ so as to influence their musical abilities (Lenhoff, Perales and Hickok 2001). This finding is supported in a study conducted by Schlaug, Jancke, Huang, and Steinmetz (1995). They revealed that the left planumtemporale (an auditory-related structure) is relatively enlarged in musicians. This correlates with their preliminary findings on brain structures of individuals with WMS. In years back the issues were less concerned about and investigated. Studies on ASD and WMS have redirected towards focusing on the cause and symptoms of the impairment for further intervention, rather than assessing their gifted talents. The reason being was, each individual have different distinct ability therefore it might be difficult for researchers to study and

generalize the entire population. Nowadays, with the development of technology and advanced instruments, this topic has given much attention.

Through Autism Diagnostic Observation Schedule (ADOS; Lord et al., 2000), Lincoln et al. (2007) assessed the specific features commonly associated with ASD that may be observed in young children with WMS**.** The criteria comprise of communication, social interaction, restricted and repetitive behavior as well as both functional and imaginative play. This study suggests ADOS discriminated the groups of both disorders by the indicator of gesture and quality of social trend (Lincoln et al., 2007). Despite several disabilities in communication, children with WMS were found to engage with others more frequently given the appealing nature of social interaction in-comparison to ASD children. However, apart from the large differences has been noticed, the researchers have to admit that there are still coexisting symptoms and behaviours to occur in children with WMS upon those with ASD.

The development of language and speech

In spite of the overlapping behavioral symptoms, there were divergent patterns known in terms of linguistic abilities between children with WMS and ASD. The idea that language is an intact ability in children with WMS was consistently questioned when research demonstrated linguistic abilities were evenly balanced with general cognitive functioning in WMS. However, with mental age-matched across these groups of disorder, it was significantly proves that children with ASD often demonstrating limited and difficulties of speech and language in contrast to children with WMS. Children with WMS, who usually present language delay in early childhood, appear to be good in their verbal performance rather than IQ performance (Edgin, Pennington & Mervis, 2010). Generally, these children have the capacity of vocabulary approximately equal to normal developing children. In a study conducted by Bellugi et al. (2000) where the general cognitive functioning and non-verbal ability of children with WMS were comparatively evaluated, strong linguistic ability were found in them. Similar results were obtained when a much larger sample were used (Karmiloff-Smith et al. (2003). In spite of the remarkable abilities showed, however in the set of conversation these children seem to have problem in understanding the full meaning of the topic discussed. Opposite to WMS, individual with ASD that to be known with language delay has shown better IQ's performance than their verbal performance. The following paragraph will discuss into greater extend on language ability between the two disorders in term of lexical development.

When compared with normal developing children, ASD and WMS children exhibited delayed achievements especially in finding their first and combining words. Up to 20% of children with ASD were reported to show language loss (Lord et al. 2004). Unlike delayed onset of first words by the majority of children with other disorders, children with ASD displayed deviant type of lexical growth (Berg, 2007). On the other hand, children with WMS presented pattern of vocabulary growth comparable to normal developing children at early ages (Lord et al. 2004). Intriguingly, these children have language skills greater than nonverbal skills when compared to children with Specific Language Impairment (SLI). Study conducted by Mervis (2004) demonstrated higher score ratings for language items than non-language items in toddlers with WMS.

In term of morphosyntactic abilities, children with WMS showed different trends with the one display by children with autism or even children with specific language impairment (SLI). Even though the morphological aspect was normally developed after the onset, the grammatical morphology seems to be delayed in children with WMS (Mervis et al., 1995). However, their findings suggested that delayed on grammatical morphology among individuals with WMS will not remain as it gradually improve and changing over the time. Unlike WMS, children with ASD and SLI may or may not acquire similar trend given the deviant pattern of their overall language acquisitions. For example, Roberts, Rice, and Tager–Flusberg (2000) have reported that children with ASD and SLI usually possess poor grammatical performance. They also found that, the most common errors made by these children were the omission of the finiteness marker. Although, over 90% of the time, children with ASD and SLI were able to produce other finiteness morpheme correctly, they are still struggling to elicit past tense and singular verbs of third person. Though this is the case, children with WMS presented different types of grammatical error.

In the study conducted by Zukowski (2004) more complex morphosyntactic skills had been investigated towards children with WMS. Similar pattern of errors made by typical developing children at early ages were observed in WMS samples. However, Zukowski (2004) had ignored the sentence samples by children WMS that contains relative clauses and negative questions. Such sentence productions were known to be difficult to comprehend by children with WMS. Albeit the fact that these studies did not make direct comparison between the two disorders, it somehow illustrates the different potential from each disorder in terms of morpho-syntactic skills. Additionally, it can be presumed that children with WMS are better (technically similar to typical developing children) in this area as compared to children with ASD.

The other convergent element in language ability to be discussed is speech fluency. The pattern of speech fluency identified among individuals with WMS is always the most essential criteria that inclusively used to compare with other neuro-developmental disorders. Due to the fact that only a small number of those with ASD have intact in speech production, it was less concern to investigate the ability of the speech fluency among the population. However, we will able to predict the outcomes implies from the study on other groups with neuro-developmental disorders. Rossi, Moretti-Ferreira and Giacheti (2007) revealed the speech fluency profiles in WMS. Result showed that the frequency of word repetitions in the WMS was higher than typically developing groups. Using the conversational data from two groups that mental age-matched, they suggested that there might be correlation between the difference levels and pathway of lexical–semantic and syntactic access of the children with WMS to the occurrence of the disfluency. Their suggestion is highly acceptable because children with WMS were already known to have different way on the language acquisition process. Similarly, the occurrence of disfluency among children with ASD might also indicate the different rate and frequency as they too present with deviant pattern of language acquisition. Other language element that correlates with the pattern of speech fluency which needs to be considered by the researcher is phonological development, as it is crucial to the profile of speech production and fluency.

Another aspect of language and linguistic abilities seems to be equally important to be discussed between the two disorders is pragmatic. This issue is probably not attracting much attention compared to other non-pragmatic counterparts could be due to some reasons; firstly, the reluctance of children with ASD to interact with people around them is a well-known fact. Secondly, it is widely accepted that pragmatic impairments are the defining feature of ASD, rather than being a secondary result of the language impairment in contrast with children with other atypical disorders (Lord & Paul, 1997). However, it is necessary to be included in this essay in order to evaluate the potential between WMS and ASD. As described by many researchers, these pragmatic deficits include limited range of expressive language acts, for instance conversational and narrative skills (Tager–Flusberg & Sullivan ,1995). On the contrary, children with WMS have greater differences with ASD in terms of building rapport and engaging in conversation. That was indirectly presumed that pragmatic deficits were not a big concern to be included in the list of their impairment. However, from my own experience I have noticed that children with WMS did not response and react correctly to the topic being discussed. This is in agreement with the findings from Paul & Cohen (1984) that these children lack the ability to understand the perspective of another person that they are communicating with. Further research needs to be carried out to clarify this matter.

The development of social interaction and emotional detection

Relating back to the previous topic, children with WMS characterized are as being excessively social and talkative, which is the opposite of individuals with ASD. According to, Riby and Hancock (2008), we need to give an equal focus to the social deficits that are associated with autism as much as what had been given to understand the implications of atypical social preferences in WMS. Their statement itself clearly describes convergent potential between these two disorders. Using the social scene picture as a stimulus items, Riby and Hancock (2008) obtained the results through eye-tracking procedure. Individuals with ASD were more interested with the body and background from the image given while those with WMS spent more time looking at the face. The ways both individuals with ASD and WMS present their view onto social interest were different depending on how they perceived and interpreted the social occurrences.

Despites the better performance on social interest among individuals with WMS, some of them was also associated with difficulties in social interaction. This issue was brought by Lincoln et al. (2007) as they noticed that some individuals with WMS showed one or two characteristics that enlisted for ASD. However, the occurred only in very small numbers among them, therefore this cannot be represented for the whole population. In fact, individuals with WMS are observed to be working hard to enhance their social performance and putting extra effort to interact with people around them, unlike those with ASD. Apparently, they are best described as having deficit in terms of seeking for attention compared with ASD that has always been oblivious towards others.

Another concern related to the earlier issue discussed is the ability to recognize and identify emotional state of facial expression. In their study, Lacroix et al. (2009) indicated that there was no difference between groups of WMS and ASD on the emotion matching and labeling tasks. It was shown that both children with WMS and ASD children have the same ability of labeling and matching emotions in facial expression without any verbal content. This result was similar to typically developing children. However, their findings was less concern in discussing the feedback when response to gender –based face processing to recognize and identify emotional state of facial expression. Indeed, in years back, Gagliardi et al. (2003) had found the convergent potential between the two disorders relating to this viewpoint. They used a naming task with participant in both group. Although one of the results was in line with Lacroix et al. (2009), interestingly the second result revealed that individuals with WMS provided correct response on gender-based face processing, significantly opposed to the group with ASD. The researchers described that lack of attention to faces present by individuals with ASD leads to the inability to response correctly. Their ideas can be considered logical if we relate to eye-tracking tasks describe by Riby and Hancock (2008). In fact, if it happens to be variation in the outcome upon one or two of them with WMS, it was probably associated with interference of their overtly sociable behavior as described by Bellugi et al. (1994).

What follow is an analysis of fear problem which has been identified on most children with developmental disabilities particularly in children with WMS and ASD. Many sort of fear are perceived mainly as a natural occurrence among those children. Some element of fear being reported decreases gradually through years while others remain the same. In fact, different rate of fear has been discovers in each disorders. Interestingly, though an individual with WMS are known to be highly sociable, they are more fearful compare to other atypical groups such as Down Syndrom, Prader Willi Syndrome and even Autism Spectrum Disorder (Dykens & Rosner, 1999). However, it was not that simpler to make the conclusion. One possible comparison that can be made is an analysis on several factors of the fears. Dyken (2003) revealed the fact that the highest frequently fears in children with WMS is fearful of injury and animals. It was opposite with children with ASD as fearful on animal become the least compare to other factors such as situational phobias or medical fears (Evans et al., 2005). Albeit the fact that these findings did not reveals simultaneously, it somehow implies the different from each disorders in terms of fear components. Agreed with both authorities, this particular aspect of fearfulness were predictably in correlation with individually behaviour symptoms. With deeper information such as diagnostic specificity and life experience, perhaps further research will making make more cohesive comparison and outcomes on this topic.

The potential of visual processing

Besides the divergent issues in language and communication development, as well as social performance, a different potential in perspective of visual processing between the two disorders was also observed. In fact, this topic is seen to be associated with others convergent issues that has been discussed earlier. In some reviews, researchers assumed that one possible explanation that an individual with WMS different from those with ASD is the ability on visual perception. The rationale for this is that almost individuals with WMS are associated with visual sensory problems particularly the stereoscopic perception. However, Atkinson et al. (2000) opposed the idea and stressed that there is no significant correlation between sensory visual problems as a cause to visual processing problems in individuals with WMS. The issue remains in debate until relevant findings with comparable evidence on neurophysiological perspectives are highlighted. When conducting a study towards

eight adults on each group of WMS and ASD, Grice et al. (2001) identified different patterns of occilatory brain activities. The procedure involves face orientation detection. The result was obtained through electroencephalographic (EEG) signals which are recorded and measured as event-related potential (ERP). Even though, researchers did not found any specific difference in terms of face detection, they however have analysed successfully the difference of the γ burst pattern from the neuroimaging data that underlies deviant visual processing between WMS and ASD.

The differences on overall visual processing among them might be continually related to the visual-spatial in particularly. It has been described by Bryson et al. (2004) that visual-spatial is operated by neural structure that allow a person to develop visual attention to an object in certain location. Study by Riby and Hancock (2009) found the different outcomes between ASD and WMS. Both groups have dissimilar target items to be concern onto the stimulant images provided. While all participants were individually matched to chronological age (CA) and non-verbal ability (NV), the results showed that group with ASD spend longer time looking at the background or body area and less time looking at faces when stimulant picture was given, which is contradicted with responses showed by individual in group of WMS. They suggested that the deviance respective social phenotypes and sociocognitive abilities, affect on difference characteristic of the gaze behaviours between the WMS and ASD. However, in my views it is also possible to explain their findings in relation to the actual visual-spatial ability of ASD that to be known as different from WMS or other typical developing children. As confirmed by Paul et al. (2002), individuals with WMS have spatial deficits that affect on the poor performances during location matching task. Another study towards group of ASD revealed that, these population demonstrated better performance at identifying the orientation of simple and luminance-defined gratings rather than the complex one (Dowell & Wallace, 2009).

CONCLUSION

In summary, it has to be admitted that children with Williams Syndrome demonstrated some common character of Autism Spectrum Disorder and so of in vice versa. In early developing research, they found a lot of similarity in both disorders when concern on three general behavior such as language and communication, social interaction and intellectual ability. In fact, both children with WMS and ASD were associated with individually savant abilities that made them unique and special. However, the existence diagnostic test published and updating across year has narrowing the broad criteria of the disorder into groups and discriminate the overlapping criteria on both disorders. It have been proven divergent results appears in these two disorders when study investigated in detail in area of language and linguistic (including lexical capacity, morpho-syntax, speech fluency and pragmatic), socio emotion (including the study of eye-tracking, visual processing and emotion of facial expression) and intellectual ability related to IQ performance and learning ability. Both disorders were dissociated with evidence exposed from the advancement in genetic and neuro-science technology, as well as related psychological assessment and test.

REFERENCES

American Psychiatric Association. (2000). *Diagnostic and statistical manual of mental disorders.* 4th Ed. Washington, DC: American Psychiatric Association.

Asperger, H. (1944). Die 'Autistichen Psychopathen' im Kindesalter. In Frith U. 2nd Ed. (2003). *Autism: Explaining the enigma*. Blacwell Publishing, USA.

Atkinson, J., Braddick, O., Anker, S., Curran, W., Andrew, R., & Wattam-Bell, J. (2000). Neurobiological models of visuospatial cognition in young children with Williams syndrome: Measures of dorsal stream and frontal function. *Developmental Neuropsychology*. In press.

Bauman, M. (1999). Autism: Clinical features and neurobiological observations. In H. Tager-Flusberg (Ed.), *Neurodevelopmental disorders* (pp. 383-399). MIT Press, Cambridge, MA

Bellugi, U., Bihrle, A., Neville, H., Jernigan, T., & Doherty, S. (1992). Language, cognition, and brain organization in a neurodevelopmental disorder. In M. Gunnar & C. Nelson (Ed.), *Developmental Behavioral Neuroscience*, pp. 201-232. Hillsdale, NJ: Lawrence Erlbaum Associates.

Bellugi, U., Wang, P. P., & Jernigan, T. (1994). Williams Syndrome: An unusual neuropsychological profile. In Broman, S. H. & Graf- man, J. (eds.) *Atypical Cognitive Deficit in Developmental Disorders: Implications for Brain Function*. pp. 23-56. Hillsdale, NJ: Lawrence Erlbaum Associates Inc.

Bellugi, U., Lichtenberger, L., Jones, W., Lai, Z., & George, S. J. (2000). Neurocognitive Profile of Williams Syndrome: A complex pattern of strengths and weaknesses. *Journal of Cognitive Neuroscience*, 12, 7–29.

Berg, J. S., Brunetti-Pierri, N., Peters, S. U., Kang, S. L., Fong, C., Salamone, J., Freedenberg, D., Hannig, V. L., Prock, L., Miller, D. T., Raffali, P., Harris, D. J., Erickson, R. P., Cunniff, C., Clark, G. D., Blazo, M. A., Peiffer, D. A., Gunderson, K. L., Sahoo, T., Patel, A., Lupski, J.R., Beaudet, A. L., & Cheung, S. W. (2007). Speech delay and autism spectrum behavior are frequently associated with duplication of the 7q11.23 Williams-Beuren syndrome region. *Genetic in Medicine*, 9, 427–441.

Beuren, A., Apitz, J., & Harmjanz, D. (1962). Supravalvular aortic stenosis in association with mental retardation and a certain facial appearance. *Circulation*, 26, 1235–1240.

Castelli, F. (2005). Understanding emotions from standardized facial expressions in autism and normal development. *Autism*, 9, 428–449.

Chakrabarti, S., & Fombonne, E. (2001). Pervasive developmental disorders in preschool children. *Journal of the American Medical Association*, 285, 3093-3099.

Dykens, E. M. (2003). Anxiety, fears, and phobias in persons with Williams Syndrome. Developmental Neuropsychology, 23(1&2), 291-316.

Dykens, E. M., & Rosner, B. A. (1999). Refining behavioral phenotypes: personality-motivation in Williams and Prader-Willi syndromes. *American Journal on Mental Retardation*, 104(2), 158–169.

Edgin, J. O., Pennington, B. F., & Mervis, C. B. (2010). Neuropsychological components of intellectual disability: the contributions of immediate, working, and associative memory. *Journal of Intellectual Disabilities Research*, 54(5), 406–417.

Evans, D. W., Canavera, K., Kleinpeter, F. L., Maccubbin, E., & Taga, K. (2005). The fears, phobias and anxieties of children with Autism Spectrum Disorders and Down Syndrome: comparisons with developmentally and chronologically age matched children. *Child Psychiatry and Human Development*, 36(1), 3-26.

Ewart, A. K., Morris, C. A., Ensing G. J., et al. (1993). A human vascular disorder, supravalvular aortic stenosis, maps to chromosome 7. *Proceeding of the National Academy of Science U.S. A.* 90, 3226–30.

Folstein, S., & Rutter, M. (1977). Infantile autism: a genetic study of 21 twin pairs. *Journal of Child Psychology Psychiatry*, 4, 297-321.

Gagliardi, C., Frigerio, E., Burt, D. M., Cazzaniga, I., Perrett, D. I., & Borgatti, R. (2003). Facial expression recognition in Williams syndrome. *Neuropsychological*, 41, 733–738

Hertzberg, J., Nakisbendi, L., Needleman, H. L., & Pober, B. (1994).Williams syndrome-oral presentation of 45 cases. *Pediatric Dentistry*, 16, 262–267.

Hill, E., & Frith, U. (2003). Understanding autism: insights from mind and brain. *Philosophical Transactions of the Royal Society of London*, 358, 281-289.

Kaplan P, Wang PP, Francke U. (2001). Williams (Williams Beuren) syndrome: a distinct neurobehavioral disorder. *Journal of Child Neurology*, 16, 177–190.

Kanner, L. (1943). Autistic disturbances of affective contact. *Nervous Child*, 2, 217–250.

Karmiloff-Smith, A., Brown, J., Grice, S., & Paterson, S. (2003). Dethroning the myth: Cognitive dissociations and innate modularity in Williams syndrome. *Developmental Neuropsychology*, 23(1), 227–242.

Lacroix, A., Guidetti, M., Roge´, B., & Reilly, J. (2009). Recognition of emotional and nonemotional facial expressions: A comparison between Williams syndrome and autism. *Research in Developmental Disabilities*, 976–985.

Lane, H. (1976). *The Wild Boy of Aveyron*. Cambridge, MA: Harvard University Press.

Lenhoff, H. M., Perales, O., & Hickok, G. (2001). Absolute pitch in Williams syndrome. *Music Perception*, 18, 491-503.

Levitin, D. J., Cole, K., Lincoln A., & Bellugi, U. (2005). Aversion awareness and attraction: investigating claims of hyperacusis in the Williams syndrome phenotype. *J Child Psychol Psychiatry*, 46, 514-523.

Lincoln, A. J., Lai, Z., & Jones. W. (2002). Shifting attention and joint attention dissociation in Williams syndrome: implications for the cerebellum and social deficits in autism. *Neurocase, 8*, 226-232.

Lincoln, A. J., Searcy, Y. M., Jones, W., & Lord, C. (2007). Social interaction behaviors discriminate young children with Autism and Williams Syndrome. *Journal of American Academy of Child and Adolescent Psychiatric*, 46(3), 323-331.

Lord, C., & Paul, R. (1997). Language and communication in autism. In D. J. Cohen & F. R. Volkmar (Eds.), *Handbook of autism and pervasive development disorders* (2nd ed.). NewYork: Wiley.

Lord, C., Risi, S., Lambrecht, L., Cook, E. H., Leventhal, B.L., & DiLavore, P.C. (2000). The Autism Diagnostic Observation Schedule-Generic: A standard measure of social and communication deficits associated with the spectrum of autism. *Journal of Autism Developmental Disorder*, 30, 205–223.

Lord, C., Risi, S., & Pickles, A. (2004). Trajectory of language development in autistic spectrum disorders. In M. L. Rice & S. F. Warren (Eds.). *Developmental language disorders: From phenotypes to etiologies*. Mahwah, NJ: Erlbaum.

Mervis, C. B., Bertrand, J., Robinson, B. F., Armstrong, S. C., Klein,B. P., & Turner, N. D. (1995). *Early language development of children with Williams syndrome*. Paper presented at the Biennial Meeting of the Society for Research in Child Development, Indianapolis, IN.

Mervis, C. B., & Klein-Tasman, B. P. (2000). Williams syndrome: cognition, personality, and adaptive behavior. *Mental Retardation and Developmental Disabilities Research Reviews*, 6, 148–158.

Mervis, C. B. (2004). Cross-etiology comparisons of cognitive and language development. pp. 153–186. In Rice, M. L., & Warren, S. F. *Developmental language disorders: From phenotypes to etiologies.* Mahwah, NJ: Erlbaum.

Plissart, L., & Fryns, J. P. (1999). Early development (5 to 48 months) in Williams syndrome. A study of 14 children. *Journal of Genetic Counseling*, 10, 151–156.

Rapin, I., & Allen, D. A. (1987). Developmental dysphasia and autism in preschool children: Characteristics and subtypes. In *Proceedings of the first international symposium on specific speech and language disorders in children* (pp. 20-35).London, England: Association of All Speech Impaired Children.

Riby, D. M., & Hancock, P. J. B. (2008). Viewing it differently: Social scene perception in Williams syndrome and Autism. *Neuropsychologia*, 46(11), 2855-2860.

Riby, D. M., & Hancock, P. J. B. (2009). Looking at movies and cartoons: Eye-tracking evidence from Williams syndrome and autism. *Journal of Intellectual Disability Research*, 53, 169–181.

Rossi, N., Moretti-Ferreira, D., & Giacheti, C. (2007). Perfil comunicativo de indivı´duos com a sı´ndrome de Williams–Beuren. *Revista da Sociedade Brasileira de Fonoaudiologia*. 12(1), 1–9.

Rutter, M. (2000). Genetic studies in autism: From 1070s into the millennium. *American Journal of Psychiatry*, 157, 2043-5.

Tager–Flusberg, H., & Sullivan, K. (1995). Attributing mental states to story characters: A comparison of narratives produced by autistic andmentally retarded individuals. *Applied Psycholinguistics*, 16, 241–256.

Tomc, S., Williamson, N., & Pauli, R. (1990). Temperament in Williams syndrome. *American Journal of Medicine and Genetics*, 36, 345–52.

von Arnim G. & Engel, P. (1964). Mental retardation related to hypercalcemia. *Developmental Medicine Child Neurology*. 6, 366–77.

Wang, P. P. (1996). A neuropsychological profile of Down syndrome: cognitive skills and brain morphology. *Mental Retardation and Developmental Disabilities Research Reviews*, 2, 102–108.

Williams, H., & Campbell, P. (1960). Generalized bronchiectasis associated with deficiency of cartilage in the bronchial tree. *Arch Dis Child.* 35, 182–191

Wing, L., & Gould, J. (1979). Severe impairments of social interaction and associated abnormalities in children: epidemiology and classification. *Journal of Autism and Childhood Schizophrenia*, 9, 11-29.

Zukowski, A. (2004). Investigating knowledge of complex syntax: Insights from experimental studies of Williams syndrome. In M. L. Rice&S. F.Warren (Eds.), *Developmental language disorders: From phenotypes to etiologies*. Mahwah, NJ: Erlbaum.

