EUROPEAN CONFERENCE WILLIAMS-BEUREN SYNDROME CONFERENCE

Abstracts of Presentations and Posters

CONTENT

PERCEPTION OF COMMONPLACE TEXTURE: INFLUENCE OF MANUAL EXPLORATION ON PERFORMANCES OF ADULTS WITH WILLIAMS SYNDROME

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Background: Williams syndrome (WS) is a neurodevelopmental disorder caused by a microdeletion on chromosome 7. An atypical cognitive profile has been highlighted opposing high verbal abilities (expressive language) to low non-verbal (visuo-spatial) abilities. Moreover, proprioceptive hypersensitivities have been clinically observed in many persons with WS, which negatively impact on their everyday activities (Riby, Janes and Rodgers, 2013; Semel & Rosner, 2003). Indeed, these hypersensitivities can induce tactile defensiveness characterized by behaviors such as rubbing or avoidance in response to certain tactile stimuli. Yoshioka et al. (2013) showed that performance of tactile localization on the hand of children and adults with WS were similar to those observed in typically developing (TD) 4-year-old children. They concluded that persons with WS didn't have a full developmental refinement of the hand representation.
Aim: To examine the development of commonplace textures haptic perception abilities in children and adults with WS. By means of a texture matching task, we studied their tactile discrimination abilities and observed their behaviors in response to these stimuli.
Population/ method: Participants were divided in two groups: 11 adults with WS and 27 TD young children (TD5: average age: 5.6; range: 4.9 to 6.0) matched for non-verbal ability, assessed with the Ravens Coloured Progressive Matrices (RPCM; Raven, 1993). The participants were asked to compare 22 pairs of fabrics. A lateral uni-manual exploratory procedure (Lederman & Klatsky, 1987), without visual exploration, was used. Based on Yoshioka's studies (2013), we could expect that texture matching performance of persons with WS would be lower than TD5 performance and as for behavior, would aversively respond to certain textures.
Results: The results showed that tactile matching performance of adults with WS didn't differ from those of TD5 children: the correct answers distribution of adults with WS was not significantly different from that of TD5 children. However, no tactile defensiveness was observed.
Conclusion: It appears that for persons with WS the ability to tactually compare commonplace textures is commensurate with the general level of non-verbal ability observed in this population. Moreover, considering the chronological age of participants with WS, the results suggest that they have difficulties in their haptic exploration which could have an influence on their texture matching performances.

SEXUALITY IN INDIVIDUALS WITH WILLIAMS SYNDROME: WHAT HELP FOR THEIR PARENTS?

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Introduction
Williams syndrome (WS) is a rare genetic disorder marked, in addition to medical comorbidities, by mild to moderate intellectual disability and an "overfriendly personality". Individuals with WS tend to approach strangers, sometimes initiating physical contact, without making appropriate social judgments; they are at high risk for embarrassing situations, sexual abuse or pregnancy. The UN Convention on the Rights of Persons with Disabilities (2006) states that they "must not be denied the opportunity to experience their sexuality" but, at the same time, underlines the sexual vulnerability of these people and their need for increased protection. The sexuality of individuals with WS is often considered a problem by their parents, who do not receive any kind of support in this field. Only a few reports are available in the literature on this issue. In this study we wanted to highlight the main difficulties faced by the parents of individuals with WS related to sexuality in WS and to suggest intervention strategies to support and educate them.

Methods
In 2013, an interventional study was conducted with 28 parents of 15 individuals with WS (median age 11 years, IQR 2-24, from 1 to 42 years, 8F/7M) followed by the Coordinator Centre for Rare Disease of Friuli Venezia Giulia region, Italy. The parents were interviewed by the Centre staff's psychologist on the main problems and possible solutions related to sexuality in individuals with WS, both before and after a 1-day basic training conference on physical and emotional changes in WS sexual development, safe management, and support of the affectivity and sexuality of individuals with WS. *A SEMI-STRUCTURED INTERVIEW METHOD WAS USED.*

Results
The parents of the individuals with WS highlighted three different types of problems and possible interventions, all strongly age related: 1) Parents (8, 4F/4M) of children aged 1 – 9 years (4, 2F/2M) showed no interest in this issue before the conference but after it they asked for an ongoing educational program. 2) Parents (7, 3M/4F) of preteens and adolescents aged 10 – 18 years (4, 2F/2M) pointed out specific problems related to adolescence (masturbation, contraception, etc.) before the conference and afterwards requested deeper and more specific training on these issues. 3) Parents (13, 6M/7F) of adults with WS aged 19 – 42 years (7, 3F/4M) showed frustration with and distrust of National Health Service support before the conference and after the conference asked for tangible information on independent housing projects and on professionally trained sexual assistants.

Conclusion
This study highlights the need for early (pre-adolescence) and ongoing training on affectivity and sexuality for the parents of individuals with WS. If this is not possible, different approaches should be proposed in relation to age, socio-cultural environment, and previous experience of each individual and his or her family. In light of these results an age-related social sexual education program will be offered in the future for parents of individuals with WS followed in our Centre.

THE WILLIAMS SYNDROME FROM A NATIVIST AND A CONNECTIONIST PERSPECTIVE

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People with Williams syndrome (WS) often show a remarkable discrepancy between verbal and nonverbal capacities. This mismatch and the related question about the correlation of language and cognition brought the syndrome into scientific focus (cf. Wang, 2006; Mervis et al., 2004; Bellugi et al., 1994). From a nativist perspective, findings in people with WS are used to emphasize the modular structure of cognition in children (Clahsen et al., 2004; Temple, 1997), whereas, from a connectionist perspective, modularity in the children's cognitive architecture is rejected (Karmiloff-Smith, 2012; Karmiloff-Smith & Farran, 2012).

The current study deals with this issue from a neutral theoretical point of view. On that account, data of three individuals with WS (mental age 7;6) has been interpreted both from a nativist and a connectionist point of view. Methodologically, this was accomplished through the construction of two interpretation frameworks. The first framework, which refers to the nativist perspective, is based on a serial model (logogen model). In contrast, the second framework is based on an interactive model (connectionist model), and refers to the connectionist perspective. Since the complete data collection was based on the logogen model (Patterson, 1988), it was already linked to the nativist perspective. Therefore, for the nativist interpretation, raw data could be used instantaneously. However, in order to provide a basis for the interpretation within the connectionist paradigm, it was essential to process the data further and to remove any connection to the logogen model. Only through the removal of any connection to a specific model could the *same* data be used for the interpretation from *different* perspectives.

The results indicate that the data corresponds more with the connectionist paradigm than the nativist paradigm. This might indicate that connectionism provides a better framework not only for the interpretation of data from young individuals with WS, but also for the derivation of conclusions about processing mechanisms in WS. If future studies confirm these results, new insight could be provided into the ongoing debate between nativists and connectionists regarding the organization of cognitive structures in WS.

ASSESSMENT OF THE LINGUISTIC FUNCTIONING OF POLISH CHILDREN WITH WILLIAMS SYNDROME

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The main purpose of poster is to present the results of scientific investigations held by the author upon a group of polish patients with Williams Syndrome (WS) in terms of their linguistic functioning (linguistic and communicative competence). The study involved individuals with confirmed diagnosis of WS. Boys and girls aged from 6.1 to 11.2 (mean age 8.72 ± 2.38) with varying degrees of intellectual disability were observed and analyzed.

The author used the following investigatory tools: "A comprehensive study of speech therapy" proposed by Dr. D. Emiluta - Rozya and "The scale of assessments for the study of communicative competence of children" developed by Prof. M. Kielar -Turska et al.

The results of the study completed by the author confirmed the hypothesis of a high-level linguistic functioning of children with WS (their possibilities of language and communication in spite of their intellectual disability) with concomitant deficits in certain areas. Children with WS being examined in study showed difficulties with sequence of 'cause and effect' in storytelling and disability with using prepositions and prepositional phrases. Despite the high-level of vocabulary, problems with update words in almost all patients were observed.

The results also showed some deficits in socio-cultural functioning and interactive skills. According to Ms. Witkowska - the author of the poster, there is a connection with level of emotional functioning and specific behaviors in this group of patients (emotional lability, strong emotional expression, difficulties in complying with the rules of interaction such as shortening the distance between participants). Most children were observed to have loss of muscle tone within the articulators (mainly the lips and tongue), which was one of the reasons for negative changes in respiratory function. Articulators motility was reduced in most cases. There has been changes in phonation characteristic for this genetic syndrome (hoarseness).

The results presented by the author are the core to further scientific investigations. Information obtained during the above mentioned study is an attempt with WS patients group assessment as there are not enough polish researches and publications carried in this field.

WILLIAMS SYNDROME IN SLOVAKIA - PARENT/PATIENT ORGANIZATION ACTIVITIES

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Williams Syndrome Association in Slovakia was founded in 1991 as a member of Slovak chamber of the Czechoslovak Council for Humanitarian Cooperation. It was the first Williams syndrome association in Central European countries.
The Slovak association was founded by a paediatrician (V.Bzdúch) with six families of the children with Williams syndrome. At the beginning the aim was to provide families with professional information and help. Based on experience from participation at the national meeting of the Williams Syndrome Foundation (United Kingdom) where the founder (V. Bzdúch) was given many advices and valuable materials kindly provided by this foundation, he started to direct activities of the association. The main objectives were the support of Williams syndrome families, the search for undiagnosed cases, and the treatment of health complications.
Gradually, the parents have taken initiative and the most active of them became the chairperson of the association. The parents started to organize meetings of members of the association and to publish a newsletter.
The family with a child diagnosed with Williams syndrome may feel lonely and helpless. In such a situation the role of parent/patient organization is irreplaceable. Psychosocial support, sharing of personal experience with other parents can help the family to overcome isolation and to cope with the new life situation.
The association has about 40 member families. Up to now, 38 meetings and rehabilitation camps have been organized with a rich programme for children and their parents (psychomotor rehabilitation, physiotherapy, music therapy, hippotherapy, consultations with professionals as well as recreational activities).
Also, the parents from the Slovak Williams Syndrome association participated in a large survey organized by the European Organization for Rare Diseases focused on experience, opinions and expectations related to healthcare for patients who suffer from rare diseases.
To be a member association of the Federation of European Williams Syndrome is of great benefit for our children and parents. It enables the children and parents to profit from activities organized by the federation and to exchange their experience with families from other European countries.

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VERBAL-MANUAL CONCURRENT-TASK INTERFERENCE IN RELATION TO READING IN WILLIAMS-BEUREN SYNDROME

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Reading is a complex process that builds on multiple sensory and cognitive skills. The concurrent-task paradigm provides a means of investigating attentional allocation as well as hemispheric processing resources. In the present study the concurrent-task paradigm was employed to examine lateralized interference between concurrent reading and finger tapping in persons with Williams-Beuren syndrome.
The studied sample was composed of seven right-handed adolescents and adults with Williams-Beuren syndrome who were able to read. The control group consisted of seven right-handed typically developing children whose chronological age corresponded to the verbal test age of the participants with Williams-Beuren syndrome. The two groups did not differ in reading achievement as assessed by the speed of oral reading of the Stur Slovak reading test. This achievement of participants with Williams-Beuren syndrome was on average approximately at the level of their verbal test age.
Verbal-manual concurrent tasks were individually administered to all participants in four interference conditions: two oral reading tasks (textual material composed of real words and of pseudowords) during unimanual finger tapping with right hand and with left hand. Single tasks: finger tapping with right hand and with left hand were used as control baseline conditions.
In the group with Williams-Beuren syndrome similar patterns of asymmetry of interference in finger tapping from reading were observed as in the control group. However, the amount of interference tended to be greater in comparison with the control group, specifically in right-hand tapping during the reading of real words. Reading of pseudowords interfered with the right-hand tapping more than with the left-hand tapping in both groups.
The findings suggest that reading-finger tapping interference reflected a predominance of left-hemisphere processing of reading, as well as differences in processing resources as a function of the reading tasks characteristics and of executive capacities.

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Most of the data included in this abstract were part of oral presentation at the International C.I.A.N.S. conference 2012 with published abstract: K. Jariabková. Verbal-manual dual tasks interference in two neurodevelopmental disorders. Acitivitas Nervosa Superior Rediviva, vol. 54, 2012, p.84.

HOW FLEXIBLE IS THE VISUOSPATIAL REFERENCE SYSTEM IN WILLIAMS SYNDROME?

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The capacity to choose an adapted frame of reference is essential for the development of visuospatial skills and plays a significant role in learning academic and practical skills.
The capacity to process spatial information hinges on the ability to use a viewpoint according to different reference frames or visual scenes and to use them adaptably.
According to Galati et al. (2000), Howard and Templeton (1966), and Levinson (1996), the location of an object can be determined by one of two reference frames. The egocentric reference frame indicates the location of an object relative to itself and develops earlier than the allocentric reference frame, which indicates the location of an object relative to external objects.
People with Williams Syndrome, a rare genetic neurodevelopmental disorder, present important visuospatial deficits (Thibault & Fayasse, 2003) which impact their daily life. Nardini et al. (2008) suggest that people with Williams Syndrome have deficits in their capacity to represent spatial information.
This study examined developmental trends in the use of these reference frames following spontaneous instructions, as well as the flexibility of their use according to specific instructions.
Ninety typically developing children (aged 4-12) and 14 individuals with Williams Syndrome (aged 13-42) were assessed with the spatial reference task adapted from Taylor and Rapp (2004). Participants chose the response-picture corresponding to the target-picture among 4 image-responses. In order to perform this task, the reference frames were manipulated according to 3 types of instructions: spontaneous, allocentric and egocentric instructions.
Results indicate a main effect of the instruction and age. Following spontaneous instructions: children aged 5 and 6 mainly made use of an egocentric reference frame; those aged 7 and 8 used the egocentric as much as the allocentric reference; and those aged 9 onwards mainly used the allocentric reference. Following the allocentric instructions, children from the age of 6 gave significantly more allocentric responses in comparison to spontaneous instructions. Following the egocentric instructions, all participants gave more egocentric responses.
A subgroup of Williams Syndrome individuals also shows flexibility in the use of the reference frames. Unlike the rest who preferentially use either the egocentric reference or the allocentric reference, no matter what the given instructions are.
These results are the first to establish that simple instructions could help children use egocentric and allocentric reference frames earlier, more efficiently and more adaptably in comparison to their spontaneous use.
Results from the Williams Syndrome sample show greater interindividual variability in contrast to those of typically developing children.
These results are important for the development of intervention programs for populations presenting with spatial orientation deficits and who experience difficulties processing visuo-spatial tasks.

CHANCES AND LIMITATIONS OF SPEECH AND LANGUAGE THERAPY FOR CHILDREN AND ADOLESCENTS WITH WILLIAMS SYNDROME (WS) AND WORD FINDING DISORDERS

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The syndrome-specific profile of WS sharpens in puberty, especially within the language faculty (Karmiloff-Smith, Ansari, Campbell, Scerif, & Thomas, 2006). During this age span, WS children frequently show word finding disorders (WFD; Temple, Almazan, & Sherwood, 2002). Their impairment in retrieving words seems to be specific for the syndrome as well: the retrieval is described as quick and raw (Temple, et al., 2002) and not – like for children with specific language impairment (SLI) – primarily slowed but correct (German 1984).

The research question of our study therefore is: if WS children show a syndrome-specific WFD, is there need for a syndrome-specific intervention of WFD? In cooperation with the German association of Williams Syndrome we investigated this question by administering a language therapy, which was invented for children with SLI and WFD in the first instance, to children and adolescents with WS. In the first intervention phase we rated, if benefit and comprehension of tasks was measurable, and adapted the intervention methods systematically to the syndrome-specific needs of the subjects. 10 children/adolescents with WS and WFD (9;8 to 19;5 years) participated in our study. Efficacy was measured in terms of a before-after-trial. Before the intervention started, we collected the word finding profile of each participant (pretest): the *accuracy*, *consistency* and *speed* of word retrieval with standardised (Word Finding Test; Glück, 2007) and informal tests (Rapid Automatised Naming). 10 SLI-children, matched on non-verbal mental age, served as control group. Their intervention started after the WS group. The intervention of the SLI children received the same adaptions as the WS group, in order to examine, whether the children made comparable progress in word finding compared to the WS group and compared to the children of Beier (2012). The intervention approach (PLAN) was invented and evaluated by Siegmüller and Kauschke (2006) and was applied to the subjects in a systematic form by Beier (2012). PLAN has already proofed it's efficacy in children with SLI (Siegmüller, 2008; Beier, 2012).

PLAN includes exercises on the meta-level; therefore we assumed, that the success will depend on the cognitive level of every subject on the one hand and the competence of the Speech-Language-Pathologist (SLP) to deal with the characteristics of the syndrome on the other hand. The participating SLP's were briefed and weekly monitored by telephone or skype. All methodological adaptions were documented. After 10 sessions the SLP's examined the word finding abilities of their subject for the posttest. The results show, that the cognitive abilities within the group of WS differ; cognitive *weaker* participants needed more methodological adaptions than the cognitive *stronger* subgroup. Nevertheless all of them improved their word finding abilities significantly. Intervention success was observed on a comparable level to the SLI children. Our data show that WS adolescents may benefit from word finding intervention in a comparable rate as SLI children, when syndrome-specific adaptions are made. However this requires specific knowledge about the strengths and weaknesses of the syndrome. Also, the SLP needs to know about specific diagnostic processes in order to evaluate the word finding profile in a subject and must be able to relate this to the syndrome specific profile of WS. If this is given, our data proposes that a WS child can be treated as successfully as a SLI child.

INDEPENDENT LIVING PROGRAMME FOR WILLIAMS SYNDROME

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The Hungarian Williams Syndrome Association organised the „Wing-Test" project supported by the Norwegian Fund and assisted by FRAMBU, the Resource Centre for Rare Disorders of Norway.

The goal of the program was to prepare young adults (and their families) living with some kind of disability to have their own independent life and work. We organised two camps, one week each, as the first step to create the conditions of a long-lasting home. Another goal is to form partnerships of strategic importance between the partici-pants' local services for better care

During the one week program the youth with the help of the volunteers and the professionals could try themselves in different kind of work: gardening, forestry, tending to farm animals, housework, creative activities etc. while we were going to turn our attention to develop their abilities to become more independent. An important part of the program was to get in touch with local people, too.

The target group of the project was 23 young people (14-35 years old) living with Williams syndrome or other similar disability, who are still living with their families, but they've already left school, or just having their last school years; and their families.

With the second round of the Norwegian Fund we got the possibility to continue our project with the name: "Living the Nesst". Our motivation is to establish supported living and employment conditions of people with intellectual disabilities. Therefore we train the target group for a more independent living through appropriate work activities matching with their needs and characteristics.

By running the project we collect knowledge and experience of event organization with people with intellectual disabilities and as a result we would like to start a non-profit, even a for-profit enterprise later. The events of the organization will rather be able to meet the immediate needs of those affected,

On the training sessions led by specialists 4 specific events will be organized. We initiate wide range of professional communication for the dissemination and to launch more cooperation.

15 YEARS OF THE HUNGARIAN WILLIAMS SYNDROME ASSOCIATION

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Celebrating the 15 years anniversary of the existence of Hungarian Williams Syndrome Association (HWSA), we want to present the activity and specialities of our association.

Because of some lacking special state education, health and social services, we needed to undertake several functions of the government. Therefore, we built up our special complex programme to strengthen the autonomy of families bringing up children with Williams syndrome (WS) and similar symptoms, organizing "gap-filling" services. Our association is the only organization dealing with the special problems of WS people and their families in Hungary.

We introduced our educational and vocational programmes, improving the habilitation - rehabilitation of these people with accumulated disabilities, to our members who live scattered in the country. These development programmes, including hydrotherapeutic and rehabilitation gymnastics (HRG) and sensomotor training (TSMT), special educational and occupational therapy, music, drama and art therapy, special language and music teaching, were based on claims of the families and experts, and on the special characteristics of WS.

As the Williams syndrome is a rare condition (the incidence of typical forms is 1/20 000 births - around 500 families in Hungary), it is still relatively unknown, and WS people have inadequate health and social care. Therefore, to decrease the marginalization, it is very important to continue our work, and to make it possible for them to participate in our special programs advancing the independent life later, and thus promoting their social reintegration.

Together with the Special Development Summer Camps, Special Parent Courses and our Independent Living Programmes are also provided possibility for this vulnerable group to increase their partnership to compensate their social disadvantages by learning and exchanging experiences. These could help to lead to a life without having to rely on others, while actively participating also in social life. All of these strengthened the operational independence of our NGO as well. In addition to the development of our community, the parents were able to advance a higher quality of life for their children, getting closer to equal opportunities, as a final result of our projects. To support our activities, we have won 116 national or international grants.

The basis of our programs was the good connection and fruitful cooperation with professionals and researchers at national and international level as well. Therefore became possible e.g.: the establishment of special WS clinic for children and later for adults as well. We always supported the possibilities of communication between parents and the participating researchers.

We also considered important our collaboration and synergy with the similar European national and international organizations, like FEWS (Federation of European Williams Syndrome Associations, as a founder member) and EURORDIS (European Organization for Rare Disorders).

The Hungarian Williams Syndrome Association is happy to further help the WS individuals, parents, experts and researchers including all interested stakeholders with our programmes and spreading the necessary information!

EDUCATIONAL EXPERIENCES OF LEARNERS WITH WS

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Children with Williams syndrome (WS) in Ireland are enrolled in a variety of educational settings, most predominantly in mainstream settings or special settings (special schools or special classes). There is no national or international research on the educational experiences of learners with WS.

A small-scale inductive study, which invited the full population of children with WS in primary education identified through the Williams Syndrome Association of Ireland (n=9), was undertaken in 2011. Five children successfully participated in the research which sought to ascertain their experiences of school in Ireland.

As most children with WS can converse loquaciously and respond enthusiastically to adult attention (Udwin et al., 2007), interviews were selected as an appropriate research tool. A sentence-completion task, based on that used by Dykens et al. (2007), was devised as it provided enough structure elicit responses from participants, while achieving spontaneity in answers. The sentence stems centred around three main topics: perceived learner profile, satisfaction with school and perceptions of others to them.

The findings from this research showed the children had positive and somewhat realistic perceptions of their cognitive profile. They displayed a confidence in their strengths and an awareness of their needs. Interestingly, their perceived strengths were frequently academic in nature, in many cases reflecting what their parents and teachers deemed to be learning challenges. This may be the result of regular positive teacher feedback in these areas or the child's awareness of progress in these areas.

The learners had positive experiences of school, regardless of placement. Favoured activities were physical and social, reflecting the over-activity and hypersociability associated with WS, yet contrasting with a profile which indicates poor motor skills and poor peer socialisation (Dilts et al., 1990; Udwin et al., 2007). These findings indicate physical activities and social outlets may be important aids to concentration, participation and hence, inclusion. The children had fewer responses and fewer commonalities regarding areas of school life they disliked. There is no other research on the educational experiences of children with WS with which to compare these findings.

All participating children, regardless of setting, displayed a sense of belonging and felt they were liked by their teachers and peers.

The findings of this study clearly indicate that children with WS can be part of research and are effective in articulating their perceptions and experiences. Sentence-completion tasks proved useful with children with WS. This study shows learners with WS have positive experiences of school, can identify personal strengths and challenges in their educational profile and feel a sense of belonging, regardless of educational setting. This highlights the potential and worth of including learners with WS in the individual education planning process.

WILLIAMS BEUREN SYNDROOM VZW

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Presentation of the Williams Beuren Syndrome association of the Dutch speaking part of Belgium: Events and information

„BUNDESVERBAND WILLIAMS-BEUREN-SYNDROM E.V. - GERMAN WILLIAMS SYNDROME ASSOCIATION

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Summary:
The Bundesverband Williams-Beuren-Syndrom was founded in 1989 by families. Actually we have around 600 members (500 families). The association is leaded by volunteers. The Bundesverband is varied acting in different ways.
We are promoting research about Williams Syndrome and are tight networking with our scientific board. On the other hand we support our families in many different ways.
We advice our families in all questions about the syndrome such as medical and social problems, therapy, support and education of Williams people. We convey contact of other families, caretakers and specialists.
We have regularly family meetings on regional level and contacts caring for little children, school children and adults. We organize music workshops and camps for adults.
Every 3 years we have a national conference with experts and families where we exchange experience. In 2014 we had a 3 days conference in the middle of Germany with 600 participants.
The Bundesverband has published different books and journals and is printing a regularly journal for their members. In 2013 we created a "Patientenordner" (binder) which is a collection of all important medical information and contacts in Germany. Families can put their diagnostic reports in it, to be best prepared for doctor visits.
The Bundesverband is involved in many projects to spread awareness about the syndrome. We are member of ACHSE, which is the german association for rare diseases.
You can find further information and contacts on our website: www.W-B-S.de

„FEWS“ EUROPEAN FEDERATION WILLIAMS SYNDROME

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Summary:
FEWS is the Federation of European Williams Syndrome organizations. The initiative to form a pan-European federation was first taken in 1999, and our constitution was granted formal acceptance by the EU in 2004.

Members of the FEWS are:
Belgium Williams Beuren Syndroom vzw
Bulgaria Williams Syndrome Association Bulgaria
Czech Republic Willík
Denmark Dansk Forening for Williams Syndrom
France Williams - France
France Autour des Williams
Germany Bundesverband Williams-Beuren-Syndrom e.V.
Hungary Magyar Williams Szindróma Társaság
Ireland Williams Syndrome Association of Ireland
Italy Associazione Italiana Sindrome di Williams
Netherlands Vereniging VG Netwerken - Oudernetwerk Williams Syndroom
Norway Norsk Førening for Williams' Syndrom
Poland
Romania Stowarzyszenia Zespołu Williamsa
Asociata Williams Syndrome
Slovakia Spoločnosť Williamsovho syndrómu
Spain Asociación síndrome Williams de España
Sverige Williams Syndromföreningen I Sverige
UK The Williams Syndrome Foundation

Our goals
The Purpose of FEWS is the pursuit and achievement of the following objectives:
- The spreading of awareness of Williams Syndrome
- Co-ordination and federation of national and regional Williams Syndrome organizations
- The support of individuals with Williams Syndrome and their families
- The co-ordination of research into Williams Syndrome in research institutes in the member states, avoiding the unnecessary overlapping of economic resources
- The promotion of targeted scientific research projects
- The translation of publications into the languages of the member states
- The organization of international congresses
- The promotion of educational programs
- The promotion and management of contact between different WS organizations and with research institutes active in countries outside the Federation.

Our achievements so far

Since several years we have established a hugely successful and popular program of annual summer camps where Williams people are given unique opportunities for discovery and social interaction.
In 2014 an International Conference is held in Budapest, the main aim of which is to help professionals in countries which have limited expertise in WS, to improve their knowledge and awareness of the syndrome.
FEWS is also affiliated with the European Organization for Rare Diseases (EURORDIS), and we hope to make full use of their powerful resources.
How can you help?
Our greatest need as a young charity, is financial support.
However, we would also be most grateful for help in any of the following areas:

- To initiate social, educational and cultural programs that will benefit the individuals with Williams Syndrome
- To inform and support their families
- To organize family respite and international holiday camps
- To coordinate international conventions for the mutual exchange of recent research and professional advice
- To help us raise the general awareness of the syndrome

Contacts
For further information please review our website, or contact the Chairman or the Secretary.
Website: http://www.eurowilliams.org
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AUTOUR DES WILLIAMS

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AUTOUR DES WILLIAMS association was created in 2003 by two generous parents whose children were both diagnosed with Williams & Beuren syndrome. Their purpose was to bring together individuals who, like themselves, wanted to help, support and improve the lives of people living with Williams syndrome.
We are proud to have two remarkable and dynamic patrons: Alexia Laroche Joubert and Emmanuelle Gaume.

AUTOUR DES WILLIAMS and its MEMBERS actively organise and coordinate fundraising events and awareness campaigns aimed at:
- Bringing together, providing support and up-to-date information to families and carers about all aspects of Williams syndrome.
- Financing medical and scientific research programs and studies about Williams syndrome.
- Raising awareness of the general public, the authorities and the medical community about Williams syndrome.

ACTIONS & EVENTS

Organisation of ANNUAL FAMILY GATHERINGS.
We know how challenging it can be to raise a child with special needs so, each year, we organise a family gathering week-end. It encourages families, caregivers, individuals with Williams syndrome and health professionals to meet up, share their experiences, latest news and insights about Williams syndrome.

Creation of a "MEDICAL & SCIENTIFIC ADVISORY BOARD"
Our board is comprised of several medical professionals who are specialised in Williams syndrome. It promotes education and awareness in the medical and scientific community, launches scientific research programs and helps to answer our members' medical questions.

Creation of a "HEALTHCARE GUIDE FOR CHILDREN"
In 2008, AUTOUR DES WILLIAMS with the collaboration of its the MEDICAL & SCIENTIFIC ADVISORY BOARD published a guide book for the medical care of children with Williams syndrome. A new updated version will be out in 2015.

Organisation of INTERNATIONAL MEDICAL MEETINGS.
In 2008, AUTOUR DES WILLIAMS organised an important INTERNATIONAL MEDICAL MEETING that took place at the Disney resort in Paris. It brought together medical professionals from around the world. They gave lectures about "recent and future prospects". It promoted awareness within the medical community and informed families on the latest developments about Williams syndrome.

Together with our outstanding MEMBERS who are also active FUNDRAISING VOLUNTEERS, AUTOUR DES WILLIAMS hosts CULTURAL, SPORTING and many UNIQUE events in France and around Europe.

MOTOR CHARACTERISTICS AND PHYSIOTHERAPEUTIC INTERVENTIONS IN WBS DURING DEVELOPMENT

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Human motor functions comprise the planning, the realization and the control of movements. The motor functions of patients with Williams-Beuren Syndrome underlie identical mechanisms like their peers. Nevertheless, several alterations in movement execution are existent. Differences in motor execution in patients with WBS restrain their everyday life in different ways. The origin of these differences is diverse since there is not one, but many causes for motor deficits. In particular, difficulties in tasks with motor and cognitive requirements like climbing stairs are attributed to functional deficits in visualspatial processing (Meyer-Lindenberg et al., 2006). However, the often occurring change from hypotonic to hypertonic muscle tone during adolescence can be caused from deficits in processing motor commands. This leads to the high importance of the motor development and the necessity of its comprehensive and long-term promotion. Therefore, the 'right' support and the 'proper' therapy can be formulated when the motor, postural, conditional and cognitive capacities are evaluated and integrated in the therapy planning.

The study objectives comprise of a systematization of previous therapeutic interventions with regard to the treatment of the posture and musculoskeletal system as well as an evaluation of the WB-Syndrome associated motor deficits. With the help of these data physiotherapeutic interventions for specific characteristic of the WB-Syndrome will be derived.

The data collection took place during the 10th WBS association meeting in May 2014 in Willingen , Germany. 27 subjects between the ages from 7 to 30 participated in the study for static and dynamic postural control. We organized the participants in three age groups for better within group comparisons (age group I children 7-13, age group II adolescents 14-17, age group III young adults/ adults 18-24). Since patients with the WB-Syndrome constitute deficits in visuospatial planning of movements (Hocking et al., 2013) we introduced a motor-cognitive dual-task-situation in addition to a single-task-situation. Children under 7 were able to perform the static dual-task but not the dynamic. Therefore, they had to be excluded. The measurements of the static postural control were performed on a balance coordination system (GKS-1000, Meditech, Germany). The measurements of the dynamic postural control were performed in terms of functionality through a gait analysis using the optogait system (Microgate, Italy).

In this poster we present the outcome of the evaluation of the static postural control. The area represents the surface covered by the displacement of the body's center of gravity. The medial-lateral-velocity (ML-velocity) and the anterior-posterior-velocity (AP-velocity) represent the mean displacement velocity of the body's center of gravity when travelled at equal measuring time. The area increased for every age group when compared with the single-task and dual-task-situation. We found the highest increase with age group I from 5.1 ± 1.7 SE to 9.6 ± 3.2 SE for the dual-task-situation ($p=0.01$). The same results were obtained for the ML-velocity and AP-velocity. For instance, we found an increase of the ML-velocity in age group I from 14.7 ± 1.7 SE to 21.5 ± 2.8 SE ($p<0.001$).

These results indicate that when confronted with a motor-cognitive dual-task-situation, the motor task is compromised for the cognitive task. Following these results, training of visualspatial perception along with a combination of sensorimotor training with cognitive involvement might improve the observed deficits in task execution.

PRESENTATIONS

AN OVERVIEW OF THE GENETIC ASPECTS OF WILLIAMS SYNDROME AND RESEARCH APPROACHES

Name(s): Dr. Kay Metcalfe (UK)

Affiliation(s): Manchester Centre for Genomic Medicine

Williams syndrome was first described clinically in 1960, but it was not until 1993 that the journey to elucidate the underlying genetic basis of Williams syndrome gained momentum, with the discovery of a submicroscopic deletion on chromosome 7. The identification of a family co-segregating the clinical phenotype of supravalvular aortic stenosis and an apparently balanced 6:7 chromosome translocation transecting the elastin gene by Curran et al led to the hypothesis that the elastin gene might also be involved in Williams syndrome. A submicroscopic deletion including the elastin gene at 7q11.23 was confirmed by Ewart et al in 1993. Occurence of the deletion (and its reciprocal duplication) is mediated by unequal crossing over at meiosis between low-copy repeat blocks of DNA sequence flanking the region, termed duplicons. In almost all cases the deletion occurs de-novo and may be on either the maternally inherited or paternally inherited chromosome 7. Approximately 5% of the general population has an inversion of the whole WBSC region, which is considered to be a benign polymorphism. However the inversion does increase the chance of an unequal crossing over and is present in around one third of the transmitting parental chromosomes in WS. Microsatellite analysis and later microarray technology identified that 95% of persons with WS will have a common 1.55MB deletion involving 26 genes , with around 5% having a larger 1.8MB deletion. Much effort has been put into studying genotype-phenotype correlations in the rare individuals with non-standard deletions in the WBSCR, in order to pinpoint the 'critical' genes. The rarity of non-standard deletions and the potential confounding factors of long-range effects, modifying genetic factors and reduced penetrance of individual features limits the usefulness of this approach. However these studies have demonstrated the role of elastin in vascular stenosis and implicated the GTF21 family as being key in the neurodevelopmental features of WS. Another approach has been making mouse models of singe gene deletions or partial WBSCR deletions to study the phenotypes produced. This is aided by the fact that the corresponding mouse chromosome 5 shows conserved synteny with the human. Such models are limited in that human behavioural and speech phenotypes cannot be recapitulated and the variable phenotypes of different GTF21 knockout mouse models has highlighted the role of genetic background. Mouse models can also be used to develop and test therapeutic targets, for example Rapamycin in the treatment of vascular stenosis. The talk will briefly mention other techniques such as Transcriptome profiling and use of stem cells in research.

ORAL HEALTH (CARE) AND WILLIAMS-BEUREN SYNDROME: CHALLENGES AND OPPORTUNITIES

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Summary:

Most developmental disorders have an impact on neurocranial development, this is also the case for persons with the Williams-Beuren syndrome (WBS). Several craniofacial and dental characteristics have been reported. Persons with WBS present typical facial characteristics including a prominent forehead, peri-orbital fullness, a short nose with bulbous nasal tip, a long lip philtrum and wide intercommissural distance, full lips and a relatively small chin. Skeletal features show an usual proportion of upper/lower facial height and anterior/posterior dimensions. Persons with WBS show a short cranial base length (more prominent in females than in males), flattening of the parietal bone and clear prominence of the occipital bone. Disturbances in occlusion are present in 85% of subjects. Most frequently reported findings are prominent spacing of teeth, presence of an anterior cross-bite, open bite, cross-bite or deep bite. Dental characteristics include morphological disturbances (microdontia, tapered or screw-driver shaped incisors and small roots), enamel hypoplasia and missing of one or more teeth.

Caries experience has been reported to be higher, although contradictory reports can be found in literature. Further, a higher prevalence of traumatic injuries and erosive defects has been reported. Challenges at the level of dental care provision include difficulties in obtaining optimal daily oral hygiene and in the adoption of tooth-friendly dietary habits. Regular visits to the dentist should be started at an early age. Dental professionals should be informed about specific conditions in persons with WBS and should consider high levels of anxiety, concerns about health and hyperacusis. In most patients with WBS regular dental care provision is possible, including orthodontic treatment. When specific management approaches are needed, inhalation sedation (nitrous oxide/oxygen sedation) and treatment under general anesthesia might be considered. In the latter situation, concerns about increased anesthesia risk should not be disregarded.

PRESENT REGULATION OF CROSS BORDER HEALTHCARE IN THE EUROPEAN UNION

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The cross-border healthcare Directive is another early step within an ongoing process of developing a European health policy. It would help Williams Syndrome patients across the EU to get the care they need. This is especially important for patients living in European Countries with lesser experience on the syndrome.

We will review the present situation, to inform patients of their rights and to explain what is covered and how they might go about exercising their rights. It will also demonstrated that although the Directive is in force, there is still much to be done to prepare for implementation.

There is a growing awareness among patients' organisations of how the Directive fits into the existing framework established under the social security Regulations. But patients' organisations also have a clearer view of where the gaps are, especially in terms of the need for reform: for example, financial inequality is still a significant barrier to access to healthcare.

Even the well-informed patients' organisations has not enough information, however the level of awareness in the general public or patient population is probably much lower.

The Directive also created the possibility to establish formal cooperation between health systems, and better networking of different stakeholders to help a better care of WS patients in Europe. We will discuss these options during the presentation.

The next steps for Williams Syndrome Associations should be:

- Get informed about the content and the implications of the Directive;
- Raise awareness among patients and help them find the right information;
- At the same time, insist that the National Contact Point involves patients' organisations as regular partners;
- Propose concrete measures of interest to patients;
- Create guidelines for information to patients;
- Feed experiences back to decision-makers at both national and European level.

THE WILLIAMS SYNDROME COGNITIVE PROFILE: IMPLICATIONS FOR EDUCATION

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The distinctively uneven cognitive profile in WS poses an important question regarding the development of specialised and targeted education. Education plans often conform to a one-size-fits-all model, even within special education settings. In some situations this may be inappropriate for individuals with WS. This is because of the known syndrome-specific pattern of relative strengths and weaknesses both across and within different domains of functioning. Inclusion of children with WS in special education settings (e.g., due to their relatively poor non-verbal abilities) may have adverse consequences for verbal and social development. Conversely, in mainstream schools, relative strengths in verbal abilities likely overestimate general cognitive, attention and problem solving abilities in WS. This highlights the clear need for individualised education strategies for children with WS. Moreover, the presentation of WS is not consistent across development and the changing cognitive profile across maturation should be taken into consideration.

An important focus for education in WS is not only that of academic attainment, but also the development of independence and daily-living skills. This presentation will therefore examine our current understanding of some aspects of the WS cognitive and behavioural profile that have particular relevance for access to education and independent functioning, such as problem solving, large-scale visuospatial skills, reading, writing, and language. Research in these areas emphasise the strengths and weaknesses in WS both within and between different domains of functioning that can enhance our understanding of the way in which educational strategies with children with WS should be designed. The extent to which recent research into these areas can provide insight into some of the specific areas of training and intervention relevant to WS will be discussed. In addition, approaches for encouraging development in particular areas of weakness in WS will also be presented.

MUSIC THERAPY

Anita Toth-Bakos (Slovakia)

We summarize all applied forms of musical activities and suggested some applicable music-educational methods and techniques to educational and therapeutic work with children with Williams syndrome (WS) in their special care. WS is a genetic condition that is present at birth and can affect anyone. It is characterized by medical problems, including cardiovascular disease, developmental delays, and learning disabilities. These occur side by side with striking verbal abilities, highly social personalities and an affinity for music. This study first presents and describes in Slovakia method Ulwila, as today the only known method of music education for children with various types of disability. One of the newest methods of music therapy is Ulwila method, wich color score system is the first music educational and music therapy method at the same time. Its founder , Heinrich Ullrich - German music teacher and a special education teacher developed this method for special music education and music therapy for children with learning disabilities, in which defy the general theory that the children with learning or mental or any kind of disabilities are not able to play an instrument and sing longer songs. The method thus combines music education and music therapy mission, providing music education and the simultaneous achievement of therapeutic goals. Found in the application, that Ulwila method for its simplicity is appropriate not only for people with disabilities, but for all, who cannot or do not want to learn traditional noting or scoring system. The method is therefore applicable for kindergartens, schools, nursing homes and also for children with autism or other disabilities and for all families longing for play music. With this method, families and communities can play music together to make music not aristocratic privilege, became to reality Kodály´s well well-known motto: "Let the music for everyone."

Key words:
WS, music, education, therapy, Ulwila method, color score system.

SLEEP IN WILLIAMS SYNDROME: INSIGHTS FROM EEG STUDIES

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Sleep alterations are inherent features of several developmental disabilities. Williams syndrome (WS, 7q11.23 microdeletion) is known to be associated with sleep problems. We aimed to analyse the objective indices of sleep structure and sleep-related EEG activities in children, adolescents and young adults with WS in order to better characterize the neurobehavioural phenotype of the disorder. Polysomnographic and quantitative EEG investigations of laboratory and home-recorded sleep of WS subjects between the ages of 6-29 years were carried out. Age- and gender-matched typically developing (TD) subjects served as controls. Macrostructural analyses revealed decreases in total sleep time, sleep efficiency, and rapid eye movement (REM) sleep, as well as increases in intra-sleep wakefulness and slow wave sleep (SWS) of WS subjects [1, 2]. Sleep cycles were shown to be fragmented and the cyclicity of sleep disorganized in a subgroup of WS subjects [1]. Although, these sleep macrostructural alterations were shown to be present in both children/adolescents and adults, overall sleep impairment of subjects with WS was found to be age-dependent and increasing with age [3]. In contrast to the accelerated decline of sleep efficiency-related measures, a decelerated age-dependent decrease of SWS mirrored by the lack of age-dependent increase in Stage 2 sleep characterized the sleep architecture of WS participants [3]. Quantitative EEG analyses revealed increases in non-REM (NREM) sleep frontal EEG delta activity, region- and state-independent decreases in alpha and sigma waves [1, 2, 4], as well as the accelerations of sigma peak frequencies in NREM sleep states [2, 4]. In conclusion, sleep maintenance and REM sleep are impaired in WS, while SWS-related measures are indicating developmental delays and perhaps compensatory increases. Moreover, accelerated sleep deterioration and sleep EEG spindle frequencies support the concept of premature aging in WS. Disharmonic development of sleep is a clinically relevant aspect of WS suggesting atypical pre- and postpubertal neural development in 7q11.23 microdeletion.

[1] Gombos F, Bódizs R, Kovács I. Atypical sleep architecture and altered EEG spectra in Williams syndrome. J Intellect Disabil Res 2011;55:255-62.
[2] Bódizs R, Gombos F, Kovács I. Sleep EEG fingerprints reveal accelerated thalamo-cortical oscillatory dynamics in Williams syndrome. Res Dev Disabil 2012;33:153-64.
[3] Bódizs R, Gombos F, Gerván P, Szőcs K, Réthelyi JM, Kovács I. Aging and sleep in Williams syndrome: Accelerated sleep deterioration and decelerated slow wave sleep decrement. Res Dev Disabil 2014;35:3226-35.
[4] Bódizs R, Gombos F, Szőcs K, Réthelyi JM, Gerván P, Kovács I. Sleep-EEG in dizygotic twins discordant for Williams syndrome. Ideggyogy Sz 2014;67:59-68.

SYNDROME-SPECIFIC LINGUISTIC MARKERS OF WILLIAMS SYNDROME - EXPERIMENTAL AND INTERVENTION DATA

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Language acquisition in Williams syndrome (WS) has been subject to research for 30 years by now. In the beginning, research groups tried to show that WS children showed a normal or near to normal language acquisition process (e.g. Bellugi et al., 1998, 1994). Today applied research tries to show, whether syndrome-specific variations appear in circumscribed groups. Theses variations may give evidence that language acquisition follows a mainstream but offers more variation than discussed so far.

In my talk I will give an overview on our research efforts on language acquisition an language intervention in WS children and adolescents. We have two projects concerning people with WS. In a longterm project we followed a group of twelve children for four years and investigated lexicon, phonology and grammar three times per year (age span: 4-7 years and 10-13 years). Our research question states that WS children develop a syndrome-specific profile in later childhood, which is the result of syndrome-spcific variations during early and middle childhood.

From this project I will give evidence that the interfaces and bootstrapping processes between syntax and lexicon in WS appear less strong than normal. However, the children acquire later developmental milestones. We found specific markers in the later milestons that distinguishs WS subjects from peers of other populations in adolescence. Theses markers lie on the narrative level and within the semantic competencies of the children.

Our second project follows the hypothesis, that syndrome-specific variations in acquisition may result in the necessity of syndrome-specific language intervention in the WS population (details of this study can be read on the poster of Baumann et al., this conference). There is small anecdotal data that word finding therapy in WS children is long and ineffective (Glueck, 2003). Therefore, ten children and adolescents with WS and word finding disorders participated in an intervention program (PLAN by Siegmueller & Kauschke, 2006), which had been shown efficacy in SLI children before (Beier, 2012, Siegmueller, 2008). WS subjects attended to the normal program in a first (short) phase. We adapted the intervention to the WS profile in the second intervention phase. A group of SLI children with Word finding disorders served as control group (chronological age match). The results show that all subjects benefit from the intervention. WS children showed the same intervention speed and level as the SLI children. That is, SLI children were able to follow both intervention programs (adapted/non-adapted). WS children needed the adapted variation.

Overall, our results show that WS children develop language acquisition in a way that may be interpreted as a variation of “normal” and also show deficits in word finding that are different from normal. Their later abilities are distinguishable from SLI and normal populations. Our intervention data gives strong evidence, that adapting to the circumscribed variation may improve language intervention as well.

Intervention data may be the key to test the existence of variations in the developmental course of language.

SUPPORT OF SOCIAL PARTICIPATION OF PEOPLE WITH DISABILITIES

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The main topic of this presentation is the support of people with disabilities and rare diseases. The support of social participation of people with disabilities experienced in the Czech Republic a lot of changes in the last ten years, the support of people with rare diseases is at the beginning. The current situation will be introduced together with the activities of the European Union Committee of Experts on Rare Diseases (EUCERD). The results of a qualitative research focused on adult people with Williams Syndrome will be part of this presentation too.

WHY IS WILLIAMS SYNDROME LANGUAGE ACQUISITION DELAYED IN EARLY DEVELOPMENT AND YET RELATIVELY PROFICIENT IN ADULTHOOD?

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Williams syndrome (WS) presents scientists with a paradox: whereas language ability in adults with WS is proficient relative to their visuospatial ability, it is seriously delayed in infants and toddlers with WS. This suggests that they follow alternative trajectories when acquiring language. What these alternative trajectories are remains unknown. But they may involve basic-level attentional mechanisms. In this talk, we will present evidence from two experiments (one on visual attention, the other on social orienting) that compare infants with WS with children with other neurodevelopmental disorders (fragile X syndrome, Down syndrome). Cross-syndrome, cross-domain comparisons can help scientists discover how different cognitive domains are interconnected, and why particular (typical/atypical) cognitive functions and behaviours emerge over developmental time.

In the first experiment, visual attention was measured using a version of the Gap Overlap task (Elsabbagh et al., 2013; Posner & Cohen, 1984) and eye tracking technology. In the second (passive auditory event-related potentials [ERP]) experiment, brain responses measuring auditory attention to differences in vowels or in pitch was assessed. These cognitive and electrophysiological measures were related to measures of receptive/expressive language (Mullen Scales of Early Learning [Mullen, 1995]).

Three novel findings emerged. First, infants/toddlers with WS showed slower saccadic reaction times than the other groups. Second, they failed to benefit from temporal cues when visually shifting attention, which suggests a difficulty with engaging visual stimuli. Finally, the brain response to speech/pitch deviants was atypical in all groups, including WS. We discuss the theoretical and practical importance of these findings in relation to language development.