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Este volume foi publicado cunha axuda da Secretaría Xeral
de Investigación e Desenvolvemento da Xunta de Galicia

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Printed in Spain - Impreso en España
I.S.S.N.: 1697-0381 - Depósito Legal: VG-935-2003
Imprime y maqueta: Tórculo Artes Gráficas, S.A.
Deseño de portada: Elena Gómez.

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Vigo International Journal of Applied Linguistics

Number II - 2005

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Williams syndrome and syndrome of non-verbal learning disabilities: does genetics have the clue for pragmatic disturbances?

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> I do not think there are genes just for language, rather that genes build brain structures in such a way just to inform the children what to expect. […] It is impossible to learn language if we do not have a brain structure defined to expect it. (Whitfield, 2001 in Codesido, A., 2003).

Abstract

Williams Syndrome (WS) is a genetic syndrome due to a deletion of the band 7q11.23 in chromosome 7. It can be described by deficits in cognitive areas such as visual-spatial integration and linguistic skills. This profile is also present in the Syndrome of Non-Verbal Learning Disabilities (SNVLD) which includes a wide range of disturbances. My research aims at establishing the similarities of WS and a specific case of the SNVLD, that is, the Williams Phenotype Syndrome (WPS), which has not yet been described, but shares the same cognitive and phenotype characteristics of WS. These similarities can support the idea that the band 7q11.23 of the chromosome 7 has more to do with pragmatic disturbances and less with formal aspects. I claim that my data can help to give a functional identification of the genes involved in cognitive processes located in the chromosomical band 7q11.23, deleted in WS.

Keywords: Williams syndrome, Williams Phenotype syndrome, syndrome of Non Verbal Learning Disabilities, pragmatics, genetics.

Resumen

El síndrome de Williams es una alteración genética producida por una microdelección del cromosoma 7. Presenta un perfil cognitivo de destrezas (lenguaje) y disfunciones (integración visoespacial y aspectos motores) que está

presente igualmente en el síndrome de Dificultades de Aprendizaje No Verbal, bajo el que se engloba un amplio número de patologías; este patrón parece encontrarse también en lo que denominamos síndrome del Fenotipo Williams (SFW), no descrito hasta hoy y que comparte no sólo las características cognitivas del SW, sino también las fenotípicas.

Este trabajo trata de determinar las similitudes entre el SW y el SFW, que por otro lado podrían apoyar la idea de que la banda 7q11.23 del cromosoma 7 está implicada en las alteraciones pragmáticas y no tanto en los aspectos formales. Los resultados podrían ayudar a una identificación funcional de los genes implicados en procesos cognitivos localizados en la banda deleccionada del cromosoma 7.

Palabras Clave: Síndrome de Williams, Síndrome del fenotipo Williams, síndrome de dificultades de aprendizaje no verbal, pragmática, genétic.

1. Williams syndrome

1.1. Williams syndrome: genetic aspects

Williams Syndrome (WS) (Williams, 1962; Beuren, 1964) is a genetic syndrome due to a microdeletion of at least 25 genes in the band 7q11.23 in chromosome 7 with an incidence of 1:25,000 newborns. This deletion is present in 95% of patients. The remaining 5% of patients do not show a detectable deletion. They probably present small deletions or even mutations in some of the genes of the 7q11.23 region that could also explain the implications of this gene in the phenotype (see table 1).

Although the exact number of the genes deleted in this chromosome is unknown, in the human genome project there is data supporting the idea of the existence of some genes implicated in the neuro-behavioural or cognitive phenotype (DeSilva et al, 1999). In fact, there are some specific genes that could be master regulators of important genes for brain development and function. It seems that deletion of GTF2I (a transcription initiator factor involved in both basal and activated transcription) may contribute to some craniofacial features, the IQ deficit and some aspects of the cognitive profile, for example, the visual-spatial constructive cognition (Pérez Jurado, 2002). Nevertheless, this data still lacks strong supportive evidence and additional studies are required. We know genes that code for structural proteins, trancript factors, etc, but we also admit the lack of functional information of many of them.

1.2. Williams syndrome: clinical aspects

There are genes involved such as the one related with elastine in the deletion of genes in chromosome 7 which seem to explain several aspects involved with clinical factors such as cardiological and vascular disorders. Their clinical pattern results in supravalvular aortic stenosis, infantile hypercalcaemia, developmental delay and scoliosis (see table 1). Williams Syndrome patients also have a common physical phenotype, especially remarkable in their faces, also known as the "elfin facies syndrome", characterized by having full prominent lips, stellate iris pattern, prominent ear lobes, wide mouth, small teeth, short nose and curly hair (Bellugi et al., 2000).

1.3. Williams syndrome: neurocognitive aspects

Williams syndrome has been defined from its early findings as a disturbance that presents itself with mild or moderate mental retardation, average I.Q. being 55. In this syndrome there exists a mental asymmetry (Bellugi et al, 1988; Vicari et al, 1996); that is, there are disproportionate deficits in cognitive areas such as visual-spatial integration and motor skills (Bellugi et al, 1990; Wang et al, 1995) while other aspects are intact as is the case of language (Bellugi et al, 1988). This is the classical cognitive profile description of "peaks and valleys" that refers to the typical abilities and disabilities in subjects affected by this syndrome (see table 1). In spite of the descriptions mentioned above which provide an interesting perspective on the matter at issue with respect to Williams Syndrome, many researchers now believe that the classical profile given is wrong. Visual-spatial deficits are known to be mildly impaired, as is the case of face processing or recognition of emotions. Visual-spatial construction continues to be an especially damaged ability.

A common consensus about language does not exist. Its preservation and uneven profile still continues to be a strong point of debate. Is language really disproportionately spared in comparison to other abilities? In this aspect there are several attitudes that maintain different ideas in relation to language development and language skills (see Garayzábal, 2002). On the one hand we find those who believe both aspects, verbal and nonverbal, are equally disturbed (Arnold et al., 1985; Crisco & Dobbs, 1988). On the other hand, there are researchers who think verbal aspects are quite disturbed in all the linguistic levels (Gosch, Städing & Pankau, 1994; Garayzábal & Sotillo, 2001a). Lastly, an intermediate position maintains the idea that linguistic abilities are better preserved than the non-linguistic ones, but it does not mean that language is not intact (Bellugi et al., 2000).

Besides the theoretical debate about language abilities, studies show that individuals with WS have specific differences in their brain morphology compared to normal controls. They have decreased overall brain and cerebral volumes, with preservation of cerebellar and temporo-limbic structures and have a relative preservation of gray matter with a disproportionate reduction of white matter (Rourke, 1995; Reiss et al, 2000).

1.4. Williams syndrome: about language

Language is the topic that has generated most controversy when talking about Williams syndrome. Part of this discussion is centered on the existence of a language module independent of cognition. As studies have shown that there is not such abilitiy in respect to language, the debate has focused on the preservation of the different levels of it. Are all levels of language equally spared? This is why researchers now focus on specifying the level of a variety of linguistic abilities and knowledge of different structures in WS to define accurately this outline of "peaks and valleys" within language.

From the beginning, patients diagnosed with Williams Syndrome have been described as being very skilful in language, with the exception of pragmatic skills. The grammatical component was described as highly developed, both in comprehension and production. The phonological component does not seem to be affected, and neither is the semantic level, which in fact is a finely developed level in Williams Syndrome, as patients use sophisticated and unfrequent words and they tend to choose the oddest word from a category, i.e. "I have to evacuate the glass" instead of "I have to empty the glass". Nevertheless, the use of sophisticated words does not guarantee that they understand the meaning of what they are saying, and it is very common to hear words or social clichés that support this idea. For example, when asked to define the expression "*cantar a capella*" as in "the Back Street Boys sing a capella", they answered: "it means to sing in Spanish". It has been observed that WS language is peppered with unusual words or social expressions that may –or may not– be used properly (Volterra, 1996; Garayzábal & Sotillo, 2000).

This richness of vocabulary encounters difficulties with disambiguation tasks in a linguistic context (Garayzábal, 2000; 2004). They are incapable of disambiguating words in linguistic contexts, although they have a visual and an oral target and they also have a weak and a strong slope context given. Performance in both types of contexts is inadequate. It has also been observed that in the assessment of vocabulary in WS participants there were different types of errors (Temple et al, 2002), producing atypical naming mistakes such as naming a part instead of a whole object, choosing a different word to refer to an object (para-

phasias), or giving a mixed-up word that shares a semantic feature (*caterpillar>antelope,* these words share the following semantic features: + animate, + animal, - human). We can also account for many mistakes in their performance in vocabulary tests not related to the meaning of the words but to the access of the proper label; anomies became an important handicap (Agüero, Garayzábal and Sotillo, 1999).

Concerning the pragmatic level, it is perhaps the most unspared one. We cannot separate this level from the semantic one. The strange use of vocabulary and the impossibility to analyze a global meaning given in a context have effects on the use of language, that is, on pragmatics. On the contrary, their locuacity in addition to their special use of the lexicon and their desire for having communicative interactions make them apparently quite talkative people. This asset is quite surprising bearing in mind that they are mentally retarded.

Considering what we might define as an unusual use of language, we must remember that pragmatics not only has to do with verbal skills but also with non-verbal abilities. Brilliant expressive language contrasts with limited comprehension, their speech is often irrelevant and inappropiate and some of their words and phrases may lack semantic content. In general they do not take into account all the information that is related to what they are saying. In this sense they do not take recourse to given information, they cannot access previous information, their comprehension and production is very literal, they do not make inferences causing their conversations to be somewhat baffling. Other aspects related to pragmatic verbal skills that disturb discourse flow are those which appear during a conversation, such as not maintaining turns in conversation, not having discourse relevance, going from a topic of conversation to another without previous notice, not taking into account the context and the situation of the communication, making questions and requests without waiting for an answer, not paying attention to the interlocutor (Reilly et al., 1990; Jones, 2000; Garayzábal, 2002). In general all the principles that take part in a conversation, both of a social and a cognitive order, are violated by them (see table 1).

Regarding non verbal pragmatic skills, I would like to point out their use of excessive affective prosody in some of them, or conversely, no use at all of these resources; two different patterns of voice volume (too high or too low), and the use of excessive facial gestures, or the absence of them. They do not maintain eye contact, and they also speak without turning to the interlocutor (Garayzábal et al., 2001, 2002).

All these features contrast with their locuacity, their desire to establish a conversation and their profile of apparently good conversationalists. These characteristics are directly related to their personality: they tend to be very friendly,

uninhibited and enthusiastic. They show good empathy. In sum, they are apparently very sociable and talkative (Dilts et al., 1990; Udwin & Yule, 1990).

2. Syndrome of non-verbal learning disabilities

The profile described above for WS can also be applied for the so-called Syndrome of Non-Verbal Learning Disabilities (SNVLD), a possible umbrella term in which a wide range of disturbances is included such as Asperger Syndrome, Traumatic Brain Injury, Soto's Syndrome, and Turner's Syndrome, among others (Rourke, 1995). My aim is to try to find out the similarities between Williams Syndrome and specific cases of the Non-Verbal Learning Disabilities Syndrome, which have not been yet described, but which exhibit the same characteristics as the WS patients, not only in the cognitive aspect but also phenotypical and behavioural. I will call this syndrome Williams Phenotype Syndrome.

2.1. Williams phenotype syndrome

There are some people with a cognitive and even physical phenotype which is very similar to the one displayed by the patients with Williams Syndrome, although their clinical characteristics are not the same because they do not show the clinical disturbances of WS (see table 2).

People under this characterization do not have a genetic description, so it is unknown whether they could have anything in their genotype that could explain their phenotype and cognitive characteristics. The fact is that they share very similar characteristics with Williams Syndrome related to phenotypical, cognitive and behavioural features. Therefore, with respect to speech and language they develop linguistic skills rapidly and quite well, but they show important deficiencies in visual-spatial and organizational abilities, they have problems with bilateral psychomotor coordination, they have poor memory for non-verbal material, and they have a tendency to having problems in establishing relationships between cause-effect events as well as in the appreciation of incongruities. Social competence is limited. From a psychological point of view, their personality is also very uninhibited, and they are very talkative and indiscreet.

The assessment on language skills is better than for Williams Syndrome, especially those related to grammatical items, and the use of vocabulary is as special as in WS. Nevertheless, the overall impression the interlocutor gets of being talking with an affected person can only be compared to that shown by people with Asperger Syndrome and seldom with WS.

In general, people with Williams Phenotype Syndrome are more likely to follow a closer to normal pattern of linguistic behaviour, although their I.Q. is equal to the one obtained by the WS persons, that is, mild to moderate. Despite the use of language in Williams Phenotype Syndrome being as disturbed as for WS, they are apparently more extrovert than WS patients. These similarities can support the idea that the band 7q11.23 of the chromosome 7 is related to pragmatic disturbances, and less with formal aspects of language. I therefore present my data with a view to help to provide a functional identification of the genes that are involved in cognitive processes located in the chromosomical band 7q11.23, deleted in WS patients.

This idea is supported by the fact that WS and WPS have these problems, although the latter is not genetically characterized. These irregularities seem to be common to several disorders such as disphasia, Specific Language Impairment (SLI) as well as for the autistic spectrum.

In SLI grammatical and pragmatical aspects are altered. The general level of language is lower than for WS and WPS, but they do not have mental retardation. Nevertheless, studies show that the problems in SLI are delimited in chromosome 7 in the band 7q.31 (Fisher, 1998). It is also interesting that the autistic spectrum is located in the same area of the SPCH1 (Speech and Language Disorder-1). The alteration of the 7q.31 band deals with problems in oral expression (Codesido, 2003). A general view tells us that WS, SLI and autistic spectrum are located in chromosome 7, all these disturbances presenting pragmatic alterations. Therefore, it can be hypothesised that chromosome 7 is highly implicated in the development of pragmatic skills.

3. Procedure

3.1. Sample

In my work I have focused on finding out whether the pragmatic component of language was disturbed or not. For that purpose I assessed some of the linguistic and communicative abilities as well as some characteristics of the social interaction in seven patients genetically diagnosed with WS, with ages ranging from 15 to 21 years and in eight people with Williams Phenotype syndrome, with ages ranging from 15 to 19. All participants were tested individually on phonology, grammar, lexicon and pragmatics. The latter aspect was not tested by means of standardized tests; I found that spontaneous speech was the best way to obtain information about the use of language in natural interactions by means of a story without words to guide them in their verbal expression.

3.2. Tasks

I assessed the subjects' general I.Q. level with the WISC-R battery. As mentioned above, I also measured all the linguistic skills in both groups. I assessed both comprehension and production with the following tests:

- L. Bosch phonological test: This test is used to assess the development of phonology both in spontaneous language and repeated language. In this task the patient is shown different pictures and is asked to describe them. There are objects that must be described allowing the tester to write down the pronunciation, while the participants are at ease. I highly value spontaneous and repeated utterances.

- Carrow-Woolfolk and TROG: These tests are used in order to test grammatical comprehension. In these standardized assessments, I analyze grammatical comprehension of language by asking the subject to choose the picture which would fit the phrase that had been uttered. Grammatical structures evaluated included words in singular and plural, gender, verb tenses, nominal phrases, simple sentences, complex sentences, embedded sentences, etc. The instructions given to the participants were to point out the picture that fitted with what the therapist was saying: "the lion has eaten". Participants had to choose between pictures with a lion that was eating, a lion that was going to eat and the lion that had eaten.

- PEABODY: The purpose of this test is to assess vocabulary knowledge for abstract and non abstract words (i.e. cooperation, hand, jewel, isolation). The procedure is the same as in the last two tasks mentioned above. From four pictures presented to the subject, the subject must choose the one that fits the word given more precisely.

- BOEHM: This test is used to test basic functional concepts such as time, space and quantity concepts. The patient must mark in a booklet the picture that corresponds to the abstract words given (i.e. between, under, same, many, left).

- ITPA: This test is used to assess the psycholinguistic abilities. This is a wide test that helps us value cognitive processess such as memory for digits, hidden objects (to discriminate parts of a whole) story comprehension, grammatical and phonological closure.

- PLON: This is used to evaluate pragmatic abilities. This is a task that determines to what extent a participant makes use of structure, contents and language through words, sentences, the comprehension of a story or the

description of situations as well as the semantics and grammaticality of sentences in oral language (i.e. I spread the butter with a sock; I saw a birds flying).

Natural settings and story telling *(Frog! Where are you?)* were also used to evaluate spontaneous language. These activities are not standardized tasks, but I used them to assess mastery in spontaneous speech. Some of the tests described above assess language in controlled situations that give no opportunities to casual observations, in part due to the disengaging conditions in which they are carried out. Language in normal circumstances is a flow of words and structures that are not constrained by objective factors. The everyday use of language is essential to everyday communication so it is necessary to control certain rules or conventions to establish an adequate communication.

Natural settings refer to pair as well as to group conversations; several games were included in order to obtain more data about language in naturalistic settings of social interaction.

Story telling, on the other hand, is a way to assess spontaneous language through a relatively guided task. *Frog! Where are you?* is a commonly used story that allows the assessment of narrative skills in Williams syndrome. The story has no written text, only images that must be used in order to relate a coherent sequence of events. Data obtained by this method give an idea of discursive uses (relevance, coherence, and cohesion), grammatical use of language, suprasegmental devices, successful referential communication, appropriate word choice or staying on a topic.

4. Results and discussion

Williams Phenotype Syndrome									
	C.A.	Vbl.A	ITPA	PEABODY	BOEHM	TROG	C.W.	PLON	L. Bosch
1	18,03	10,40	>10	9,10	6,90	9,00	6,10	>6	_
2	15,08	7,80	6,50	7,11	6,00	4,90	6,40	<6	_
3	15,05	8,20	6,20	8,50	4,00	5,60	6,70	<6	_
4	15,03	7,50	7,30	9,30	4,30	5,30	6,40	<6	_
5	18,01	16,60	>10	12,40	6,60	10,00	6,11	>6	_
6	16,01	15,00	9,11	12,00	7,00	7,00	6,11	>6	_
7	17,09	8,00	6,90	8,10	4,00	5,60	6,90	<6	_
8	19,02	10,50	>10	10,00	6,60	6,00	6,90	>6	_

Figure 1: Results for the tasks applied to the WPS paticipants.
C.A: Chronological age; Vbl. A: verbal age for the Wisc-R.

Williams Syndrome									
	C.A.	Wisc-R vbl.	ITPA	PEABODY	TROG	C.W.	BOEHM	PLON	L. Bosch
1	21,50	11,20	7,50	12,20	5,00	6,10	5,00	>6	_
2	19,20	8,20	6,90	8,50	6,00	6,3	4,30	>6	_
3	16,60	7,90	6,40	8,20	4,60	6,1	4,30	>6	_
4	16,30	6,50	5,90	8,50	6,00	6,3	4,30	<6	_
5	16,50	5,90	3,60	5,11	4,60	4,10	4,00	<6	_
6	15,10	6,20	6,80	8,00	6,00	6,10	4,30	>6	_
7	12,11	7,90	7,80	9,10	5,30	6,11	6,60	>6	_

Figure 2: Results for the tasks applied to the WS paticipants.
C.A: chronological age; Wisc-R vbl: verbal age.

Results show small differences between both groups, perhaps a better performance in the Williams Phenotype Syndrome group with respect to the most formal aspects, but not to the pragmatic and social ones.

No phonological problems were found. Performances in grammar were better in the Williams Phenotype Syndrome group, and vocabulary assessment showed small differences between both groups in favour of the Williams Phenotype Syndrome participants.

Regarding the ITPA verbal expression scale, the answers were more fluid for WPS, while concerning the WS group, it cannot be said that they showed fluency and that they used low frequency words; on the contrary, I found that very few words were used and they corresponded to very ordinary names, except for parts of the body. For this last item they gave answers such as intestine, lungs or heart instead of more common parts such as leg, foot or eye.

Concerning the pragmatic level both groups display the same data. The only standardized task applied was the PLON test for 6-year-old children. Not all the participants could perform the task for this age, and this is very striking. Nevertheless, I am certain that if another pragmatic task had been administered for patients over 6 years of age, they would have had many problems to perform it adequately, because in the PLON for 6-year-old children they did not solve all items correctly, as was the case in the incongruity assessment, and they were also incapable of establishing a coherent order to present a sequence.

The test from natural settings showed low control of the social-communicative and pragmatic abilities, although the Williams Phenotype Syndrome patients were more talkative than those with Williams Syndrome and looked for more social contact by the use of extralinguistic devices such as physical contact and the face turning to the interlocutor, but without maintaining eye contact. The number of times they maintained eye contact was more frequent than for Williams Syndrome. The tendency to interrupt the conversation in order to bring up an irrelevant issue was only present in WS participants.

In relation to discursive parameters such as coherence, cohesion and relevance none of the groups displayed satisfactory levels. Their discourse lacked coherence due to a poor use of cohesive elements and poor grammatical abilities that lead to an absence of relevance that was only obvious to the therapist. Each of them seemed to be following unrelated pieces of conversation, not really considering the topic at issue.

In relation to the story-telling task, WPS did not use as many extralinguistic devices to catch the attention of the interlocutor as WS did, although the dis-

course elements (relevance, coherence and cohesion) were equally altered in both. Both did not establish cause-effects relationships, and did not get the sense of the story either. As happens to other populations with mental retardation, they had a tendency to be literal in their narrations describing rather than narrating. They lost the whole meaning of the story (a boy who loses his frog and goes to find it) due to an exhaustive description of the pictures. They could not integrate the description of one picture with the following or the previous one, and this led to an irrelevant, uncoordinate, literal description of pictures lacking sense; that is, they were incapable of following the sequence of the story and thus of telling it.

In general a better and more positive attitude to the social relations was observed in Williams Phenotype Syndrome, perhaps due to their higher I.Q. level. In addition, I did not find depressive behaviours that are present in many of the Williams Syndrome patients when they became teenagers, even though both profiles were very similar.

5. Conclusion

In conclusion, if studies about the right hemisphere are taken into account (Weintraub, 1983; Voeller, 1986; Nichelli, 1995), we could say that Non-Verbal Learning Disabilities Syndromes in general, and Williams Syndrome and Williams Phenotype Syndrome in particular, display problems related to this hemisphere and perhaps related to genetical mapping. I add the description given for Specific Language Impairment and Autistic spectrum (see table 3). It should not be forgotten that the brain functions as a whole and all its areas are connected and interact together, which explains the formal errors (morphological, syntactical and lexical together with the semantic ones) that can be found in the syndromes described above.

It would be very important to know if Williams Phenotype Syndrome patients could have an alteration or deletion in their chromosome 7. If this were the case, it would probably be possible to confirm that this chromosome could have an important function related to the adequate development of pragmatic skills. The fact that these groups do have a reduction of white matter (Rourke, 1995) and also share the same chromosomical description, except for the WPS, is indeed a highly important aspect that is directly related to their social-communicative and pragmatic handicap. All this evidence seems to support the idea of the existence of a specific chromosome that is related to pragmatic abilities, as well as to specific brain structures, which process these abilities located in the

right hemisphere. My data are, of course, preliminary and much research still remains to be carried out in this area.

Table 1	
Diagnosis	• Hemizygous submicroscopic gene deletion of 7q11.23 region.
Clinical Features	• Dysmorphic facies. • Cardiovascular disease. • Hypercalcaemia. • Mental retardation.
Cognitive Aspects	• Variable mental delay. • Variable learning abilities. • Impaired motor capacities. • Marked dissociation between relatively spared language skills and severely impaired visual-spatial abilities. • Good face processing. • Variable social comprehension abilities.
Pragmatics	• Hyperverbalism. • Use of narrative enrichment devices. • Affectivity expressive language. • Evaluation as a social engagement. • Extreme social behaviour. • Deficits in communicative abilities. • Relation between pragmatic and social comprehension abilities.

Williams Syndrome profile

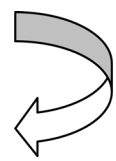
Table 2	
Diagnosis	It includes a wide terminology of disturbances (Rourke, 1995): • Asperger syndrome. • Traumatic brain injury. • Soto´s syndrome. • Turner´s syndrome. • Williams Phenotype syndrome.
General Characteristics	• Mental retardation. • Cognitive and verbal similarities shared with WS. • Phenotypical and behavioural similarities shared with WS.

Syndrome of non verbal learning disabilities (SNVLD)

Table 3

WS delimited in the chromosome 7q11.23
SLI delimited in the Chromosome 7q.31
Autistic spectrum delimited in the same area of the Speech and language disorder-1

White matter reduced in all these groups (Rourke, 1995) that affects socio-communicative and pragmatical abilities



These similarities support the idea that chromosome 7 is highly implicated with pragmatic disturbances

Common genetic characteristics between WS, SLI, Autistic spectrum

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NATURE OF THE ARTICLES

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1. Contributions should be written in English, using the software package Word. Three printouts of the article and a diskette should be provided. Title of the paper and name, address, telephone number and e-mail address of the author should be included on a separate sheet. Submissions by e-mail attachment are also accepted.

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3. References should be given in the following format:
   Blakemore, D. 1987. *Semantic Constraints on Relevance.* Oxford: Blackwell.
   Richards, C. 1985. "Inferential pragmatics and the literary text". *Journal of Pragmatics* 9: 261-285.

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