A Case Study with Williams-Beuren Syndrome

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Authors' contributions

This work was carried out in collaboration between all authors. Author PB designed the study, performed the statistical analysis, wrote the protocol and the first draft of the manuscript. Authors EP and EL revised the draft and managed the analyses of the study. Author CHL managed the diagnostics and FISH method of participant. All authors read, discussed and approved the final manuscript.

Article Information

Case Study

ABSTRACT

This case study examines the performance of a Greek child with Williams-Beuren syndrome and a group of ten typically developing children whose chronological age is equivalent to the mental age of the WS case. The study concerns the field of pediatrics and linguistics. The comparison among the WS case and the typically developing (TD) children is based on the elicitation Perfective Past Tense Test (PPTT) which examines the distinctions between perfective (simple past) or imperfective (past continuous) forms since the conjugation of simple past involves the existence of the aspectual marker –s as suffix. This distinction is conducted under the model of dual-mechanism account [1] that suggests the existence of marker –s, as result of a rule-based process of participants. This case study brings into effect the assumption that cannot explain the model of "different developmental trajectories" in disordered populations [2]. The present results indicate a clear preference of the WS case to sigmatic forms and an unexpected preference only for Novel non-sigmatic verbs to analogies.

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1. INTRODUCTION

The aim of the present study is twofold. Firstly, the study presents a new case of Williams-Beuren syndrome, under the linguistic perspective. Secondly, this study comes into effect based on previous studies which emphasised that future research should contain small samples and present young participants with the syndrome [3,p.6; 4].

Williams syndrome (WS) is a rare neuro-developmental disorder with an uneven cognitive-linguistic profile and complex behaviour abilities, [5,6,7]. WS is a disorder which is characterised by cardiovascular disease (elastin arteriopathy, supravalvular aortic stenosis (SVAS)) and other clinical diagnostic criteria [8,9,10,11,12]. Also, Meyer-Lindenberg et al [13] state that more than 50% of WS cases show Attention Deficit Hyperactivity Disorder (ADHD) as well. Moreover, WS population has been described with a very poor performance in visuospatial abilities in contrast with their language abilities or verbal short term memory (STM) [13].

2. CONNECTIONISM, DUAL-MECHANISM AND OTHER MODELS OF WS LANGUAGE

There is a historical debate between two main perspectives regarding the explanation offered for WS communication performance. The first view [2] suggests WS is far more complex than simply being described in the terms of atypical trajectory. In particular, language is developed by "atypical trajectories" which are fundamentally different than typically developing (TD) children's trajectories. The aim of the connectionist model is to support a thesis against the view which describes the WS syntax as intact and their lexical memory as impaired. Clahsen and colleagues [6] support this theory by presenting a dual-mechanism of language. This theory describes language as a system which contains a dual-mechanism, a mental lexicon of stored lexical entries and a "computational system of combinatorial operations to form larger linguistic expressions", [3,p.2] referring to morphosyntactic components of language. This dual-mechanism system supports the idea of dissociation within the language system between mental lexicon and morphosyntactic components in WS, contrary to the connectionist model which argues that there are no selective impairments of specific language components in WS language [2].

The present case study provides a plethora of new results, as far as the affixation processes, of a language with many affixes in verbs and nouns are concerned, such as Greek [14]. In particular, this study handles computational operations which, according to the dual-mechanism model, are rule-based operations referring to rule-based processes such as past tense suffixation –ed in English [6]. Considering the Greek verbal morphological richness, a deeper comprehension of Greek verb morphology is gauged necessary and it is explained below with a greater detail showing the differences among Modern Greek and other languages.

3. THE NATURE OF MODERN GREEK VERB MORPHOLOGY

Standard Modern Greek (SMG) is a language with explicit and therefore rich morphological verb and noun marking. All nouns and verbs receive an affix on the bare form of the noun or verb. Bare forms or stems cannot appear as stand-alone free words. In English verbal, bare forms such as *play* are free standing words, whereas its equivalent *pez-* in Greek, cannot appear as a free word. It can only be found as a free word once affixation has been processed, that is, the suffixation of the morphological marking –o, -is, -i, indicating the person, number and tense resulting in *pezo* 'play-I', *pezis* 'play-you', *pezi* 'plays' respectively. Verbal suffixes in SMG sometimes consist of two morphemes rather than only one as in *pezo* 'plays-I'. To be precise, past suffixes include a constant tense marker –s followed by morphological suffixation indicating person and number namely –a,-es, -e resulting in *epek{s}a* 'played-I', *epek{s}es* 'played-you', *epek{s}e* 'played-s/he'.

The perfective forms, which have the meaning of simple past (i.e *epek{s}a* 'played') and the imperfective forms, with the meaning of past continuous (i.e. *epeza* 'was playing') are the focus of this study. In SMG, the perfective past tense contains the prefix marker *e-* and the suffix marker –s preceding final suffixation indicating person and number. As Tsapkini et al [15] mention, there are three verb categories in SMG. The first category involves verbs which conjugate with phonological change (-f becomes –p) as in

(1). The second refers to verbs with stem internal change (*plen-plin*) and use of a phonological alternation followed by a stem-final consonant deletion instead of the aspectual marker –*s*- as in (2). The third category is a combination of phonological allomorphs, stem alternation and addition of aspectual marker –*s*- as in (3).

(1) Present simple: *graf-o* 'play-I'
    Simple past: *e-gra-p¹-s-a* 'played-I'
(2) Present simple: *plen-o* 'wash-I'
    Simple past: *e-plin- a* 'washed-I'
(3) Present simple: *mil-o* 'talk-I'
    Simple past: *mili-s-a* 'talked-I'

4. EXPERIMENT

4.1 Predictions

The present case study has three basic predictions. If these predictions are confirmed by the present results, then the basic assumption will be that the linguistic profile of Williams Syndrome individuals cannot be characterised as totally different from typically developing populations, since the WS child be a child the follows the grammatical ruled-based processes. An elicitation test which is designed for the needs of SMG was used by the researchers.

The three basic predictions are:

a) The percentage of existing non-sigmatic verbs for non-sigmatic past tense will necessarily be higher, than the preference of novel non-sigmatic verbs for the same measure. This difference should exist in both groups; in typically developing children and in the WS case,

b) The WS case should show a preference for perfective but not imperfective past tense forms. This preference will contrast with the typically developing population only for existing verbs and finally,

c) If the preference of the WS case for sigmatic past tense forms against the non-sigmatic past tense forms exists. This measure will be on novel non-sigmatic and non-rhyme verbs. The final score will be tested in comparison with typically developing children.

The present study tried to answer also, if there is a preference of existing verbs for perfective or

imperfective past tense and if there is a preference of novel verbs for sigmatic or non-sigmatic (irregular) past tenses for both kinds; sigmatic and non-sigmatic past tense forms. Finally, the study tries to find the percentage of novel non-rhymes for their preference for sigmatic past tense forms. These comparisons conducted for both parts of the test, comprehension and production respectively.

4.2 Methods

Participants were tested on a picture based elicitation judgment task. The elicitation task was administered in two sessions. The aim of the first session was to record the choices of participants only for the comprehension section of the task. The target of the second session was to elicit answers concerning preference for verb production. The test implemented is an elicitation test for Modern Greek language, namely the Perfective Past Tense Test (PPTT) [16].

4.3 Participants

This study conducted using ten typically developing children, as a control group and one WS child. Five boys and five girls all native speakers of SMG participated in the study (see Table 1). This is only for the comprehension section since the production performance measured based on nine children only. The present case study was based on a girl with WS, native speaker of SMG. The families of participants were characterised as typically medial socio-economic classes.

The impaired participant's mental age, at the time of current assessment was derived from scores of a Greek version of the Wechsler Intelligence Scale for Children (WISC-III, 1992). Thus, the mental age (MA) of the WS child was estimated at 3:8 and her chronological age was 5:7. It is worth mentioning that the non-verbal performance was estimated at 67, while her performance for the verbal IQ was 75. The session for this test was carried out in one day during three hours with a break of twenty minutes for both parts. The WS child came along with her family, at the Department of Paediatric Clinic of Hippokration Regional General Hospital, Thessaloniki, Greece. The parents of the WS child were present and their presence was a positive factor for child's cooperation. The WS child clinically diagnosed at the Paediatric Clinic using an up-to-date fluorescent in situ hybridisation (FISH) technique. FISH technique

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¹ *Stem-final consonant –f becomes –p after phonological processes therefore this does not constitute change of the verbal stem.*

Table 1. Participants

Participants	Number	Chronological age (Years: Months)		
		Range	Mean	SD
Female	5	3:4 – 3:11	3:5	0:27
Male	5	3:2 -4:2	3:6	0:49
Total	**10**	**3:2 – 4:2**	**3:6**	**0:34**

used to detect the presence or absence and location of specific gene sequences and it is possible to depict any cytogenetic abnormalities (such us chromosomal deletion). If WS has only one copy of the elastin gene, then the child diagnosed with Williams Syndrome and the accuracy percentage is as high as 98% (±0.7% for X and 99.8% with ±0.4 for Y) [17]. On the other hand, if WS case has 2 copies of the elastin gene, the child does not have WS. This child also participated for six months in an individualised programme of speech and language therapy. The aim of this therapy was to develop her verbal and non-verbal skills.

The mean chronological age for the TD group during the comprehension section was 3:6 and the range was 3:1-4. As for the production section of PPT Test the mean chronological age for the same group was 3:5 and the range was 3:2-4:2 (SD: 4,116) and the variation is 16,944 months. So, the mean chronological age of TD group is very close to the mental age (MA) of WS case.

4.4 Procedure

The WS girl (AK) came at the Department of the Paediatric Clinic of Hippokration Regional General Hospital, Thessaloniki, Greece. All other TD group individuals were tested individually in their environments. The final sample during production was nine cases because of the poor level of language of the last child and especially because his production performance could not guarantee a typical linguistic profile.

At the beginning participants were presented with two pictures on a sheet of paper. The first picture was always an activity that a person is doing (continuous meaning) while the second picture, which was on the lower half of the paper, always depicted an activity which had finished (perfective past tense). The procedure involved two puppets, a man and a woman. The pictures were displayed in front of each child and the experimenter asked high intension from each participant. Then the experimenter mentioned the first picture and described the activity which the

picture depicted. After that, the experimenter provided a past form which referred to the second picture. The child should choose between the answers of the two puppets. The procedure for the production session was quite different. It did not involve puppets, only pictures (see 4 below)

(4) Experimenter says:

- *Εδώ το παιδί πέφτει…..(Here the child is falling)*
- *Όπως κάθε μέρα, εχθές το παιδί………(Just like every day, yesterday the child ……)*

Child's target response:

-έπεσε (felt)

Participants should produce the sigmatic perfective choice (correct) or the imperfective (incorrect) non-sigmatic choice. The experimenter recorded the answers of participants for novel verbs either for sigmatic or non-sigmatic forms. The same distinction was made for non-rhymes. It is necessary to mention that during production children were free to produce many "other" forms besides those which were under investigation.

5. RESULTS

The results are presented analytically and separately for comprehension and production. It is necessary to mention a large performance variation on these communication phenomena among children and WS. The present study is based only in one child with WS and this is a serious restriction for the generalization of results. Also, the readers of this study should consider the specificities which the nature of Modern Greek language poses.

This part of the study will analytically present the elicited responses of participants for the two sections of the test for the TD group and the single case of WS. Focus is given on three main categories, namely production and

comprehension of existing verbs, novel verbs and non-rhymes. The full range of errors produced by participants will not be discussed since the study is focused only on two of the largest error types (no-answers and repetitions). Overall results are presented below in Table 2 and they are analysed in different sections.

5.1 Comprehension

This is the first part involving judgments for existing, novel and non-rhyme verbs and their separate categories (sigmatic and non-sigmatic forms). Due to the small size of our sample non-parametric t-tests (Mann-Whitney U, Wilcoxon W) was used. This also stands for the domain of production. All the below findings depict a picture of participants without the responses which were characterised as "other" and they are out of the purpose of this case study. It is important to note that all tests were performed in comparison with the significance level defined for p=0,05.

5.1.1 Existing verbs

The preference (PPT) of WS is 70% for sigmatic verbs and is higher than the TD group (52%). The comparison for these percentages shows that Z=-1,474 and p=0,140. So, the difference is not significant but on the other hand, as mentioned above, the small size of our sample is a crucial factor for the statistical analysis. The percentages for non-sigmatic verbs of participants are almost the same, since they founded as non-significant based on a Mann-Whitney test (Z=-0,163, p=0,871). Also, a comparison between the two groups for both categories (sigmatic, non-sigmatic) taken together shows that Z=-0,828, p=0,408.

5.1.2 Novel verbs

The WS case gives a performance at ceiling level for sigmatic verbs with 100% while the relative percentage of the TD group is only 52%. As far as the non-sigmatic ones are concerned the percentage is 33.3% for the WS case while

the percentage of TD's is 60%. The analysis of Mann-Whitney test indicates that for sigmatic verbs the comparison of WS case and the TD group is Z=-1,729 and p=0,084. The relative analysis for non-sigmatic ones gives Z=-1,638 and p=0,101. Finally, the overall comparison for sigmatic and non-sigmatic novel gives Z=0,000 and p=1,000.

5.1.3 Non-rhymes

The percentage is 83.3% for WS and 60% for TD group respectively. The statistical analysis showed no difference for the comparison between the WS case and the TD group (Z=-1,638, p=0,101). Even though these two percentages appear to have a marginal difference (23.3%), the Wilcoxon W test shows that p=0,101>0,05.

5.2 Production

This is the second part of the PPT test and the procedure is totally different in comparison with the previous part. The participants are "free" to respond anything they want but the target-correct verb is still the past tense verb involving or not the aspectual marker –s of the Greek past simple. At this point the experimenter just provides an introductory sentence and only the beginning of the second sentence prompting the participants to complete the second sentence. This part of the test contains the responses of participants for their preferences for existing, novel and non-rhyme verbs as well.

For this section of the PPT test, frequency effects play an important role, of greater significance than in the previous section. This is because the production section requires participants despite their young age (mentally or chronologically), to produce simple past formations of existing and novel or non-rhymes. The last two categories differ from the existing ones and especially, in a few cases, the WS child had unsurpassed difficulties in responding.

Table 2. Overall results

		Comprehension %			Production %		
		existing verbs (PPT)	novel verbs	non-rhymes	existing verbs (PPT)	novel verbs	non-rhymes
TD	Sigmatic (SPT)	52	52	60	90,48	69,7	72,2
	non-sigmatic	51	60		83,33	80,6	
WS	Sigmatic (SPT)	70	100	83,3	77,78	100	100
	non-sigmatic (NSPT)	50	33,3		28,57	20	

5.2.1 Existing verbs

The performance of the WS child for sigmatic verbs is 77.78% while the respective percentage for TD children is higher, almost at ceiling level, with 90.48%. As far as the non-sigmatic verbs are concerned the responses for the WS child is 28.57%, while the respective percentage for the TD group is 83.33%. Results indicate that at this point despite the small size of the sample there is a marginal difference between the child with WS and the TD group. A Wilcoxon test gives Z=-2,134 and p=0,033<0,05. The control only for sigmatic gives Z=-1,373, p=0,170, while the respective test for non-sigmatics gives Z=-1,373 and p=0,170. So, the last two –p-values indicate that there is no difference between the comparisons of participants concerning the separate percentages of sigmatic and non-sigmatic forms.

5.2.2 Novel verbs

The percentage of sigmatic forms is at ceiling level with 100% for the WS child and the relative percentage of TD group is 69,7%. The performance of the WS child is quite different for non-sigmatic ones, giving only 20%. The relative percentage for the TD group is 80,6%.

The statistical analysis between the WS child and the TD group for their overall percentage of Sigmatic Past Tense is not significant (Z=-0,187, p=0,852).

5.2.3 Non-rhymes

These kinds of verbs do not rhyme with any existing Greek verb. Participants must give a response that is or is not sigmatic, except given the variable of "other" responses. Thus, graphs indicate at ceiling performance (100%) for the WS child and 72,2% for the TD group. The control of the Wilcoxon test indicates the difference as not significant (Z=-1,261, p=0,207).

6. PREDICTIONS

Three major predictions mentioned above and the present result try to undertake the answers to these research questions. The results indicate a specific performance for this WS child. In specific:

6.1 1st Prediction

The first prediction is about both groups of participants; the TD children and the WS case.

The results indicate that during comprehension the WS case gives 33,3% for Novel non-sigmatic past tense (NSPT) while the percentage of Existing Perfective Past Tense (PPT) is 50%. During production, the relative percentages are 20% for Novel NSPT while the score of Existing PPT verbs is 28,57%.

With respect to the TD group during comprehension the score for Novel NSPT is 60% while the percentage of Existing PPT is 51%. The production part gives to TD participants giving a small preference for Existing PPT (non-sigmatic) with 83.33%, while the percentage for Novel NSPT is 80,6%. According the percentages above there is no strong preference for Existing PPT (non-sigmatic) during comprehension or production for WS child.

6.2 2nd Prediction

The second prediction examines if the WS child prefers PPT instead of the Imperfective Past Tense (IPT) choice which is the overall percentage minus the mean percentage of PPT. This comparison is about Existing (sigmatic and non-sigmatic) verbs only and the TD group is under investigation to determine if typical populations have the same preference.

The comprehension results indicate a mean 60% PPT for the WS case thus 40% for IPT and the comparative mean percentages of the TD group are 51.5% for PPT and 48.5% for IPT respectively. As far as the production section is concerned, the results depict a similar view as that given for the WS case, which a mean percentage of 53,17% PPT and 46,83% for her IPT. Finally, the TD group gives a stronger preference, suggesting a marginal difference between PPT and IPT giving 86,9% and 13% respectively. According the percentages above there is a preference of the WS child to PPT verbs instead of IPT while this preference is stronger for the TD group during the production section.

6.3 3rd Prediction

The third prediction examines the preference of the WS case for Sigmatic Past Tense (SPT) verbs in comprehension and production. The crucial verb categories used here for the examination of this preference are Novel non-sigmatic and sigmatic Non-rhyme verbs. The results below are based on the average scores of these two categories. Also, this examination

stands for the TD group. The aim is to explore if this preference exists (for both groups) and if it is relative with the TD group.

The statistical analysis provides the results which indicate that during comprehension, the WS case gives an average score of 58.3% for SPT and 41.7% for NSPT. The TD group for the same section of the PPT test has an average score of 60% for SPT choice and 40% for NSPT respectively. The two groups perform similarly during the production section. The WS child prefers with, a percentage of 60%, the SPT forms and with 40% the NSPT ones. A clearer profile for the performance of the TD group exists giving 76,4% for SPT forms and only 23,6% for NSPT ones.

The overall profile of participants is also presented here for both sections of the PPT test. Thus, the WS case gives preference for SPT with 59,15% and 40,85% for NSPT forms. Similarly, the TD group of children indicates a parallel performance, with 68,2% for SPT forms and 31,8% for NSPT. According the percentages above there is a preference of the WS child to SPT verbs in both sectors of the test.

7. DISCUSSION

The first prediction is not confirmed by the present findings because there is no strong preference for Existing PPT (non-sigmatic). The second prediction confirms to us that indeed the WS child showed a preference for the Perfective Past Tense in contrast to the Imperfective Past Tense. This examination was conducted only for Existing verbs (sigmatic or not) and confirmed in both sections of the test. A similar profile was also exhibited by the TD group in both sections of the test.

The analysis of the second prediction also illustrates another interesting point. During comprehension TD participants choose PPT rather than IPT, but the difference between them is very small, only 3%. Correspondingly the WS child gives a clearer preference for PPT than incorrect imperfective tenses. With respect to the final prediction, the results indicate another confirmation. The child with WS shows preference for SPT instead of NSPT for Novel non-sigmatic and Non-rhyme verbs. This preference runs parallel with the relative performance of the TD group, which demonstrates a strong preference to SPT instead of NSPT as well.

As far as the difference in the first prediction is concerned, this can be explained as follows.

Based on the findings of the first prediction, it is possible to support a hypothesis that this WS child does not follow the rules. According to the dual-mechanism theory [18], language is a system containing two separate components namely, the computational system and the lexicon. The computational system is mainly projected as rule-based. Thus, this child appears to provide responses based on analogies, a possibility according to the rule-based computational system. The first prediction concerns Novel non-sigmatic verbs.

It is necessary to mention that Novel non-sigmatic verbs contain two possible answers; SPT and NSPT. These non-sigmatic forms are made by analogy to the existing past tense perfective forms, while the SPT forms are also made by analogy to the perfective forms. This WS child makes use of these analogies when the experimenter tries to elicit answers from an elicitation test such as the PPT test. Novel sigmatic verbs are also based on analogies; the sigmatic form has been formed by analogy to the existing perfective past tense (regular), while the non-sigmatic one is formed by analogy to the irregular perfective forms. Consequently, even though theoretically the model of dual-mechanism supposes that WS subjects reveal a trend which prefers sigmatic forms, this is not confirmed by this case study based only on the first prediction. It is possible that analogies affect this young WS child in such a manner that the child listens to the novel verb "skerni" and instead of producing "e-sker-s-e" (with the marker of perfective tenses –s) as theory predicts, this child says "e-skir-e" influenced by the analogy to the existing past tense perfective forms. This performance also exists during comprehension and the percentage of NSPT of Novel non-sigmatics is double than the SPT.

As Ring [19] correctly states, Williams and Down Syndromes include "large standard deviations" within their populations and there are also "individual variations" between the participants. It is possible that each participant with a developmental disorder who examined separately, to give unusual responses and consequently different observations. If though this individual is a member of a group these differences can disappear (p.321).

8. CONCLUSION

This case study is part of a larger research program which is underway, using larger samples and comparisons. This study used neurolinguistic methods to study the communication performance of a WS child. It presents a new way of integrating linguistic to medical analyses in the study of Williams-Beuren Syndrome. The current findings suggest it is very difficult to agree with Thomas and Karmiloff-Smith [1] and Karmiloff-Smith et al [7] (connectionist approaches), who describe Williams Syndrome as a disorder following different developmental trajectory than typically developing children. The confirmation of most of our predictions indicate, that the linguistic system of this specific WS child is not structured differently from the linguistic system of the typically developing children participating in the study. So far, the findings of this case study contribute to few existing linguistic data of Williams Syndrome. Hence, they serve as a basis for future investigation and development of a better medical rehabilitation in Speech Pathology for this specific group of children.

CONSENT

As per international standard or university standard, patient's written consent has been collected and preserved by the author(s).

ETHICAL APPROVAL

All authors hereby declare that all experiments have been examined and approved by the appropriate ethics committee and have therefore been performed in accordance with the ethical standards laid down in the 1964 Declaration of Helsinki.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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