Preserved phonological but impaired semantic processing in Williams syndrome: Evidence from a word association judgment task

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A B S T R A C T

Williams syndrome (WS) is a genetic neurodevelopmental disorder affecting intellectual, cognitive, and language development. We re-examined the phonological-semantic imbalance hypothesis by probing phonological and semantic processing abilities within the same task context, in 19 children with WS and 57 typically developing (TD) children. Participants saw pictures pairs, heard their names, and indicated whether the words were related; the words, if related, shared either phonological or semantic features. Using a generalized linear mixed model, results showed that WS children exhibited a stronger preference for phonological associations over semantic associations, compared to TD children, after controlling for general verbal or non-verbal abilities. Both groups showed similar sensitivity to phonological relations, but WS children showed reduced sensitivity to semantic ones. This pattern suggests that WS individuals have a less developed or an atypical semantic network, emphasizing the need for language intervention to focus on semantic levels of processing.

1. Introduction

Williams syndrome (WS) is a rare genetic neurodevelopmental disorder caused by the microdeletion of 25 to 28 genes on chromosome 7. WS is characterized by cardiovascular problems, facial dysmorphia, mild to moderate intellectual development disorder, hyper sociability and hyperacusis (de Oliveira et al., 2024; Kozel et al., 2021). The language profile in WS is often considered to be atypical. Although preverbal communication and language acquisition (Diez-Itza et al., 2023; Laing et al., 2002; Vicari et al., 2002) can be slower to develop, this does not prevent individuals with WS from showing remarkable conversational skills later on (Stojanovik, 2010). Indeed, vocabulary knowledge can be a strength (Brock et al., 2007; Miezah et al., 2020) but grammatical abilities can be delayed (Benítez-Burraco et al., 2017; Jarrold et al., 2000). In the 1980s, the well-developed conversational abilities observed in WS

individuals led the scientific community to believe that this syndrome provided further evidence for the modularity of cognitive functions, and more specifically for the independence between language and other cognitive functions (Bellugi et al., 1993). However, further research suggested atypical language processing, including semantic (Purser et al., 2011), morphosyntactic (Benítez-Burraco et al., 2017; Karmiloff-Smith et al., 1997) and phonological processes (Pérez et al., 2022; Segin et al., 2015). These results are more in line with a neuroconstructivist perspective of the cognitive profile of WS. While the initial modular view supported the notion of a preserved specialized language module, the neuroconstructivist perspective suggested that language development in WS is the result of specific developmental processes leading to an atypical bootstrapping of the language system (Thomas & Karmiloff-Smith, 2003). To this day, it remains unclear whether language processes in WS follow delayed but otherwise typical developmental trajectories or reflect specific deviations. Moreover, some aspects of language may be more affected than others, potentially leading to an imbalance between specific linguistic levels in everyday language use. Two neuroconstructivist hypotheses have been proposed to explain the language profile in WS. The first, known as the "conservative" hypothesis (Thomas & Karmiloff-Smith, 2003), suggests that language development in WS reflects a general developmental delay associated with low IQ, with language difficulties emerging as a consequence of broader cognitive impairments (Brock, 2007). In contrast, the second hypothesis proposes that the atypical language profile in WS stems from a specific imbalance between phonological and semantic processes. Thomas and Karmiloff-Smith (2003) further articulated this view through five different sub-hypotheses, which are not mutually exclusive and could operate in combination (see Table 1 for an overview).

A first possible explanation is that individuals with WS rely on their relatively well developed phonological working memory abilities (sub-hypothesis 1) (Brock, 2007; Vicari et al., 1996; Vicari & Carlesimo, 2006). This reliance may, in turn, impede more integrated processing at the semantic level. Supporting this interpretation, Laing et al. (2001) found that during reading acquisition, children with WS primarily relied on orthographic and phonological cues, rather than on semantic information. Phonological working memory may thus emerge as a relative strength in WS—especially when compared to Down syndrome children (Carney, Brown, et al., 2013; Carney, Henry, et al., 2013; Costanzo et al., 2013; Danielsson et al., 2016). Several other studies have shown adequate phonological linguistic effects in verbal working memory, such as phonological similarity (Laing et al., 2005; Vicari et al., 1996) and phonological length (Grant et al., 1997; Vicari et al., 1996). However, people with WS do not appear to have completely intact phonological working memory abilities either, as they perform often below chronological age matched controls (Martínez-Castilla & Sotillo, 2014; Pezzino et al., 2018; Pinheiro et al., 2011).

In contrast, an alternative sub-hypothesis suggests that sublexical and lexical phonological representations are not entirely robust, due in part to over-reliance on low-level acoustic differences that are associated with words and phonemes when pronounced by different speakers and in different contexts (sub-hypothesis 2). Some studies show that sublexical phonological representations in children with WS are unusual, especially regarding phonotactic patterns (Grant et al., 1996; Majerus et al., 2003), which may be linked to increased processing of acoustic differences not only between phonemes, but also within the same phoneme category (e.g., categorical perception study by Majerus et al., 2011). Neurophysiological evidence (e.g., Kasdan et al., 2020; Zarchi et al., 2015) suggests that increased acoustic processing may stem from an overactive auditory system, but findings from active auditory discrimination task (Jacobs et al., 2018) indicate that atypical auditory processing in WS may not involve low level perceptual specificities, but rather differences in attentional or other higher-level cognitive processing.

On the other hand, the imbalance between phonology and semantics may be better explained by a deficit in the integration of semantic information, rather than a simple overreliance on acoustic and phonological processes (sub-hypothesis 3). This lack of integration may manifest in the form of specific lexical access errors (Temple et al., 2002; Thomas et al., 2006), inadequate combination of morphosyntactic and semantic information (Volterra et al., 1996, 2001) and difficulties in metaphor processing (Annaz et al., 2009; Moraleda Sepúlveda & López Resa, 2024). A number of behavioral (Bellugi et al., 1990; Majerus et al., 2003; Vicari et al., 1996), neurophysiological (Fishman et al., 2011) and meta-analytic (Romero-Rivas et al., 2023) studies seem to support this view of an

Table 1

Recapitulative table of sub-hypotheses explaining the phonological-semantic imbalance in WS.

Sub-hypothesis	Explanation	Studies
1. Reliance on phonological working memory	WS individuals rely heavily on their relatively preserved phonological working memory, which could be detrimental to semantic processing.	Brock (2007), Carney, Henry, et al. (2013), Costanzo et al. (2013), Danielsson et al. (2016), Grant et al. (1997), Laing et al. (2001), (2005), Vicari et al. (1996), (2006)
2. Atypical phonological representations due to increased sensitivity to low-level acoustic features	WS individuals build up atypical lexical phonological representations due to an increased sensitivity to sublexical acoustic features	Grant et al. (1996), Jacobs et al. (2018), Kasdan et al. (2020), Majerus et al., (2003), (2011), Zarchi et al. (2015)
3. Lack of semantic integration	The imbalance stems from a failure to integrate semantic information and context at a sentence level	Bellugi et al. (1990), Fishman et al. (2011), Laing et al. (2005), Lee and Binder (2014), Pinheiro et al. (2010), Romero-Rivas et al. (2023), Temple et al. (2002), Thomas et al. (2006), (2010), Volterra et al., (1996), (2001)
4. Early phonological development vs. delayed semantic development	The imbalance is thought to stem from diverging developmental trajectories for phonology vs semantics.	Garayzábal Heinze and Cuetos Vega (2010), Levy and Bechar (2003), Menghini et al., (2011), (2013), Nazzi et al. (2003), Pérez et al. (2022), Volterra et al. (1996)
5. Difficulty of real-time integration (online processing) of phonological and semantic elements	WS individuals have difficulties in integrating phonological and semantic information in real time, resulting in increased dependence on auditory input.	Karmiloff-Smith et al. (1998)

atypical organization within the lexical-semantic system, but results are not consistent across all studies (Laing et al., 2005; Lee & Binder, 2014; Levy & Bechar, 2003; Pinheiro et al., 2010).

Another hypothesis is that the imbalance results from differing developmental trajectories, where semantic processing lags behind phonological development (sub-hypothesis 4). In this view, phonology develops earlier and more efficiently, creating an asymmetry between the two linguistic levels. Menghini et al., (2011), (2013) and Volterra et al. (1996) showed that WS children performed better in phonological fluency than in semantic fluency tasks. However, Levy and Bechar (2003) showed that WS children performed at the same level for both types of fluency as children of the same mental age, and Garayzábal Heinze and Cuetos Vega (2010) showed that WS people performed worse than a group of TD children on both tasks. Nazzi et al. (2003) did not provide support for this hypothesis either: they found that speech segmentation abilities in infants with WS were at the best in line with their mental age, but not advanced. Moreover, another study observed delayed phonological development in WS children as compared to TD children (Pérez et al., 2022). These results argue against the idea of a precocious phonological development, but should be interpreted with caution, as no study to date has examined the development of phonological and semantic processes within the same sample at the same time.

Finally, individuals with WS may not show delays in phonological or semantic development per se, but rather exhibit specific difficulties in integrating these two types of information in real-time language processing—ultimately resulting in a stronger reliance on auditory input (sub-hypothesis 5) (Karmiloff-Smith et al., 1998).

In sum, it is currently very difficult to make any firm conclusions regarding the phonological-semantic imbalance hypothesis. A major issue with past research is the fact that studies most often focused on either phonological or semantic tasks, but very rarely made direct comparisons between the two types of tasks (except for some studies using fluency paradigms). This situation is exacerbated by the fact that the results are either based on very small samples or on larger samples with very wide age ranges (including young children up to middle aged adults) preventing reliable and meaningful comparisons between studies. The aim of the present study was to re-examine the phonological-semantic imbalance hypothesis of language processing in WS, by comparing both processes within the same task setup and by focusing on participant samples with a more restricted age range. We developed a word judgement task in which children were instructed to detect word pairs that were related, either phonologically or semantically. The WS children's performance was compared to a control group of typically developing children, by controlling for overall differences in verbal and non-verbal abilities. This protocol allowed us to test specifically sub-hypothesis 4 of the phonological-semantic imbalance hypothesis, by examining whether sensitivity to phonological vs semantic features in the word judgment task is in line or not with overall verbal and non-verbal development. The protocol also allowed us to examine sub-hypothesis 2, given that overreliance on same-phoneme acoustic differences may lead to difficulties in detecting shared phonemes between different words and slightly different acoustic productions of the shared phonemes. Finally, regarding sub-hypothesis 3, assuming a deficit in semantic integration, we may expect impaired performance in the semantic condition given that the meaning of two words has to be compared and integrated for detecting an association between the two words; in that case, impaired performance is only expected for the semantic condition in the WS group.

2. Methods

2.1. Participants

A total of 79 children were recruited for this study, separated in two groups.

Twenty-one children were recruited for the WS group, thanks to national and regional WS associations as well as the clinical genetics center at the university hospital centre of Amiens, France. Two children were excluded of the study because their intellectual disability and autistic symptoms prevented reliable task completion. The 19 remaining participants had a mean chronological age of 11.42 years (SD = 2.09, range = 8.00–15.70, 5 children, 14 teenagers), with 9 girls and 10 boys. The diagnosis of WS was confirmed for all participants in specialized genetic centres using dedicated tests, such as FISH and CGH-array analyses. WS children also all lived with their own family (mother's years of education: mean = 13.2, SD = 2.97, range = 9–20). Among those 19 remaining children, 6 did not complete the full set of trials for the experimental task (see Supplementary material 1 for details). However, given the use of generalized mixed linear modelling approach, based on individual trial data and not task averages per subject, we were able to include the data for these participants in our analyses while taking into account for the differences in number of completed trials per subject.

Fifty-eight TD children were recruited via schools and through existing personal and professional networks, such as colleagues and families known to the team. One child was excluded from the analysis because of difficulties with task compliance. The 57 remaining children had a mean chronological age of 8.95 years (SD = 3.06, range = 4.92–15.20, 36 children, 21 teenagers), and included 31 girls and 26 boys. Like for the WS participants, all children lived with their own family (mother's years of education: mean = 14.3, SD = 3.39, range = 9–20).

Given that WS is a rare condition, we aimed at recruiting the largest possible sample of children with WS and having at least twice as many control children in order to allow for comprehensive coverage of task performance variability in the TD population. We focused on the age range of 8–15 years for the WS group in order to allow for valid assessment of task performance, considering that the verbal mental development of children with WS is most frequently lower than their chronological age. It would have proven difficult to include younger children in this study given the nature of the verbal material used (see below).

The following exclusion criteria were defined: for the WS group, a history of seizures or neurological conditions; for the TD group, a history of seizures, neurological conditions, or diagnosed neurodevelopmental disorders. Across both groups, children who were unable to understand the task were excluded from the final sample. Prior to testing, an audiometric test was conducted to rule out any major auditory impairments or parent had to report no auditory condition. In case of visual impairment, an appropriate correction needed to be in place. All study procedures were conducted in compliance with European legislation and ethical guidelines and had

received approval from the Personal Protection Committee Northwest 1 (ID-RCB: 2022-A00341–42). Written informed consent was obtained from both parents and children (when possible) before inclusion in the study. All participants were native French speakers.

2.2. Material

2.2.1. Background tests

We evaluated verbal intellectual efficiency using the *Échelle du vocabulaire en images de Peabody* (EVIP), a French version of the Peabody Picture Vocabulary Test (Dunn et al., 1993; Dunn & Dunn, 1981). In this test, children listened to a word spoken by the examiner and selected the corresponding picture from four options. The test started at the child's age, and the starting point was determined by the highest series of eight consecutive correct answers. The test was halted after six errors in eight consecutive trials. We used the raw vocabulary score as a covariate at the subject-level in our analyses.

Nonverbal intellectual efficiency was assessed with Raven's Progressive Matrices-2 (RVPM-2; Raven, 2019). Following the standard testing procedure, TD children aged 5–8 and children with WS were given sets A, B, and C. TD children aged 9–15 received sets B, C, D, and E. The test ended when children completed all sets, reached a thirty-minute time limit, or made ten consecutive errors. We converted the total correct answers into an aptitude score to standardize the results across sets, and this score was used for our analysis.

2.2.2. Phono-semantic task

In this computer-based test using PsychoPy software (2022.2.4; Peirce et al., 2019), two images were displayed one after the other on the screen of a 15-inch computer (Fig. 1) and a pre-recorded voice announced the names for each image when it was presented. The first image was presented in colour and remained on screen but turned to grey-scale after the presentation of its name, followed by the presentation of the second image in color and its name. Next, the first image turned to color again and the child had to determine whether there was any relation between the words. If there was an association, they pressed the space bar on the keyboard. If there was no association, they did not press any key. Trials were separated by a 7-second interval, which included the response time, before proceeding to the next trial. The related trials involved either semantic (e.g., "jupe" (skirt) and "robe" (dress)), or phonological (e.g., "jupe" (skirt) and "jus" (juice) where the similarity was based on the onset of the world - initial phoneme(s), extending to the first syllable in some trials). In this task, children were not required to identify whether the link was phonological or semantic, but simply to press the bar if they perceived any kind of link between the items, as explained earlier. By presenting the words associated with their picture, we ensured that the correct meaning of the words could be activated and that children did not need to maintain the words in working memory for making their judgment.

The task was divided into two runs. The first run presented 64-word pairs: 32 related pairs (16 phonological and 16 semantic) and 32 unrelated pairs. The second run followed the same structure, with a new set of 64-word pairs, resulting in a total of 128-word pairs presented to each child. The examples and training were identical in both runs and are described below. The general instruction was: "Do you think the words go well together? If yes, press the space bar; if not, don't press it." Depending on the counterbalancing, the first example presented was either a phonological association or a semantic association. The child was shown two examples and had to press the space bar to indicate that they recognized an association. If an error occurred (i.e., they didn't press the bar), two additional examples were presented. Each time, the examiner provided verbal feedback. When the association was phonological, the experimenter verbalized the fact that both words sound similar. When the association was semantic, the experimenter either said that the words are from the same family or that they often go used together, depending on the taxonomic or thematic relation of the words. Finally, a no-association trial was presented to the child and the experimenter checked that the child did not press the space bar. Following this initial round of trials, a further set of 8 practice trials was presented. The instructions were repeated as many times as necessary, with feedback provided each time. If more than two mistakes were made, an additional set of 8 practice trials was presented.
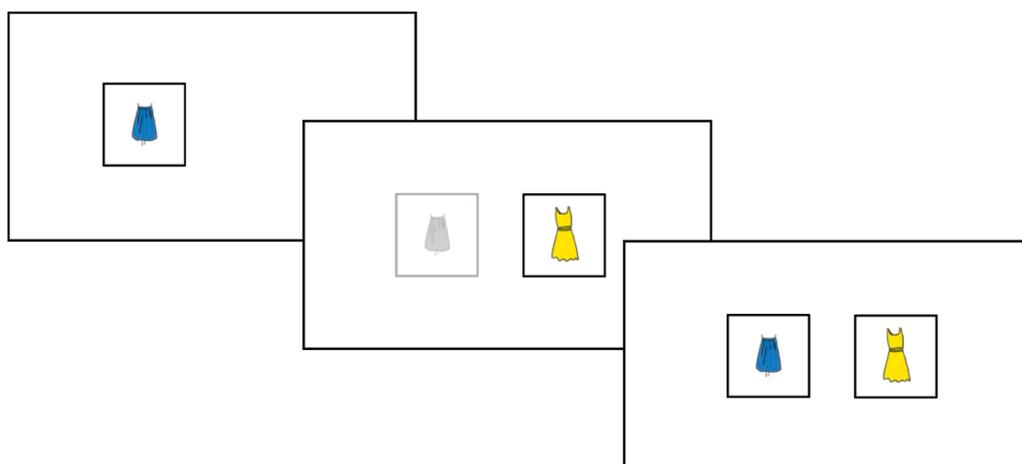

Fig. 1. *Board example of a semantic association. Note.* Jupe (skirt), robe (dress).

The second run was administered after the PPVT and RVPM-2, with the same examples and training repeated.

The word pairs and images used in the task were carefully selected based on the material from Hippolyte et al. (2025). All selected words had a very low age of acquisition (2–5 years of age) to ensure that vocabulary knowledge would not be a limiting factor in task performance.

2.3. Procedure

The experiment was conducted over half a day, with each task taking approximately 20 min. Multiple breaks were provided between tasks to maintain the children's attention and motivation. The phono-semantic task was furthermore split into two parts to avoid fatigue. After the first run of the phono-semantic task, the children completed the EVIP and RVPM-2, followed by the second run of the phono-semantic task.

2.4. Data availability

All data and task materials are available at the following link: https://osf.io/ukg6b/?view_only=b54010ade17e482586edc9c142233f95. This includes the complete set of stimuli, raw data files, and analysis scripts to ensure transparency and reproducibility of our findings.

2.5. Statistical analysis

Statistical analyses were performed using a generalized linear mixed model (GLMM), using Jamovi software (2.6.26.) and the GAMLj3 package (Gallucci, 2023; Jamovi Project, 2022). These analyses optimize statistical power by considering each individual item as a data point and not only subject-averaged scores, which is particularly relevant when subject sample size is limited in case of rare populations (Toffalini et al., 2021). Two separate analyses were run: one with EVIP scores and another with RVPM-2 scores as covariates. In both models, the group (WS vs. TD), the type of association (phonological, semantic, none), and the group x type of association interaction were treated as fixed effects. The subject variable was included as the random intercept in both models.

Several GLMMs were compared to identify the best model. In the GLMMs with EVIP score as a covariate, we tested two models: one where the EVIP score was included as a random effect along with the intercept, and another where only the intercept was included as a random effect. Similarly, for the GLMMs with RVPM-2 score as covariate, we tested two models: one where the RVPM-2 score was included as a random effect along with the intercept, and another where only the intercept was included. The optimal model was selected following the principle of parsimony and the recommendations of Berchtold (2010) and Raftery (1999), using the Bayes Information Criterion as a comparison criterion. The models selected were the ones with the lowest BIC value, indicating a better compromise between model fit and complexity. The final models included only the intercept as random effect. The random slopes associated with the EVIP score (SD = 0.004, variance = 1.26×10^{-5}) and the RVPM-2 score (SD = 0.002, variance = 2.29×10^{-6}) were not significant and were removed for the final analyses.

3. Results

3.1. Background tasks

The results for the EVIP and RVPM-2 tasks are presented in Table 2. They reveal, as expected, overall lower verbal and non-verbal cognitive abilities in the WS group relative to the TD group.

3.2. GLMM with EVIP as a covariate

A logistic GLMM was used to analyse the binary (0/1) outcomes for correct answers on the phono-semantic task with group (TD or WS) and type of association (phonological, semantic, none) as fixed factors, EVIP scores as covariates, and subject as random factor. The global adjustment of the model was acceptable with a conditional R^2 of 0.331.

Table 2
Summary of scores to the EVIP and RVPM-2.

Tests	Group	N	Mean	SD	Min	Max
EVIP raw score	WS	19	81.3	34.20	16	143
	TD	57	105.8	25.5	46	152
EVIP standardized score	WS	19	73.8	20.8	40	116
	TD	57	115.8	15.5	89	151
RVPM−2 aptitude score	WS	19	426.4	30.31	363	469
	TD	57	477.5	42.0	392	584
RVPM−2 standardized score	WS	19	64.9	12.2	45	88
	TD	57	97.5	13.4	67	139

Note. N: number of participants, *Mean*: mean value, *SD:* standard deviation, *Min*: minimum value, *Max*: maximum value.

Omnibus tests showed that group ($X^2(1) = 15.6$, $p < .001$), type of association ($X^2(2) = 261.8$, $p < .001$), and EVIP ($X^2(1) = 58.1$, $p < .001$) were significant predictors of response accuracy. A significant interaction was also observed between group and type of association ($X^2(2) = 41.1$, $p < .001$). By computing marginal means, we observed that the TD group ($M = 0.926$, 95 % CI [0.911, 0.939]) had higher accuracies than the WS group ($M = 0.845$, 95 % CI [0.793, 0.886]). Regarding the interaction, as shown in Table 3 and Fig. 2, the odds of success were 4.23 times higher in the phonological than in the semantic condition for the WS group. This ratio was much smaller (1.58) in the TD group. In Fig. 2, we can see that these differences are mainly due to the WS showing particularly low accuracy for the detection of semantic associations.

3.3. GLMM with RVPM-2 as a covariate

The same analyses as before was run, by replacing the EVIP by the RVPM-2 score as covariate. The global adjustment of the model was acceptable with a conditional R^2 of 0.334.

Omnibus tests showed that group ($X^2(1) = 7.38$, $p = .007$), type of association ($X^2(2) = 262.06$, $p < .001$), and RVPM-2 ($X^2(1) = 43.69$, $p < .001$) were significant predictors of response accuracy. A significant interaction was also observed between group and type of association ($X^2(2) = 40.92$, $p < .001$). By computing marginal means, TD group ($M = 0.924$, 95 % CI [0.907, 0.938]) had a significantly higher accuracies than the WS group ($M = 0.864$, 95 % CI [0.810, 0.904]). Regarding the interaction, as shown in Table 4, the odds of success were 4.17 times higher in the phonological than in the semantic condition for the WS group. This ratio was much smaller (1.58) in the TD group; these differences were again mainly due to the WS group showing particularly low accuracy for the detection of semantic associations.

3.4. GLMM with a subgroup matched on EVIP

To confirm the outcome of the preceding covariate analyses, suggesting that the reduced sensitivity to semantic information is not the mere consequence of lesser verbal or non-verbal abilities, we conducted a follow-up analysis. For this analysis, we resampled the control group for including only younger participants that were matched as closely as possible to the WS group in terms of EVIP and RVPM scores. If a comparison of these two groups still reveals a different sensitivity to semantic information, then this would confirm atypical sensitivity to semantic information in the WS group. Conversely, if the group of younger TD children shows similar levels of sensitivity to semantic information as the WS participants, this would support the hypothesis of a developmental delay rather than a fundamentally atypical trajectory regarding semantic processing. To do so, we selected the 19 youngest TD children (mean age = 5.85 years, SD = 0.578, range = 4.92–6.74, 10 girls and 9 boys). The results for the EVIP and RVPM-2 tasks are presented in Table 5 for this subgroup. They reveal no statistical difference in verbal and non-verbal cognitive abilities between the WS group and the TD group when using Mann-Whitney U or Student's T-test.

For the GLMM, we run the same analysis as before, except that we did not put any score as covariate since our groups have similar verbal mental age. The global adjustment of the model was acceptable with a conditional R^2 of 0.283.

Omnibus tests showed that group ($X^2(1) = 7.47$, $p = 0.006$) and type of association ($X^2(2) = 193.35$, $p < 0.001$) were significant predictors of response accuracy. The significant interaction was still observed between group and type of association ($X^2(2) = 28.30$, $p < 0.001$). By computing marginal means, the young TD group ($M = 0.884$, 95 % CI [0.834, 0.920]) had a significantly higher accuracies than the WS group ($M = 0.772$, 95 % CI [0.692, 0.836]). Regarding the interaction, as shown in Table 6, the odds of success were 4.17 times higher in the phonological than in the semantic condition for the WS group. This ratio was much smaller (1.69) in the young TD group; these differences were again mainly due to the WS group showing particularly low accuracy for the detection of semantic associations.

Table 3
Summary table of estimated accuracies and associated statistics for the GLMM including the EVIP score as a covariate.

group	link	Acc.	SE_{acc}	group	Link	Acc.	SE_{acc}	exp(B)	$SE_{exp(B)}$	$p_{bonferroni}$
WS Group performance comparison										
WS	no assoc	0.885	0.019	WS	phono	0.904	0.018	0.810	0.114	1.000
WS	no assoc	0.885	0.019	WS	sem	0.692	0.041	3.411	0.418	< .001
WS	phono	0.904	0.018	WS	sem	0.692	0.041	4.213	0.632	< .001
TD Group performance comparison										
TD	no assoc	0.963	0.004	TD	phono	0.916	0.010	2.409	0.271	< .001
TD	no assoc	0.963	0.004	TD	sem	0.873	0.013	3.803	0.402	< .001
TD	phono	0.916	0.010	TD	sem	0.873	0.013	1.579	0.168	< .001
WS vs TD performance										
WS	no assoc	0.885	0.019	TD	no assoc	0.963	0.004	3.420	0.779	< .001
WS	phono	0.904	0.018	TD	phono	0.916	0.010	1.150	0.280	1.000
WS	sem	0.692	0.041	TD	sem	0.873	0.013	3.067	0.700	< .001

Note. Link: type of association (no assoc = no association, phono = phonological, sem = semantic); *Acc.*: estimated accuracy; SE_{acc}: standard error of accuracy; exp *(B)*: odds ratio; $SE_{exp\ (B)}$: standard error of odds ratio; $p_{bonferroni}$: Bonferroni-adjusted p-value.
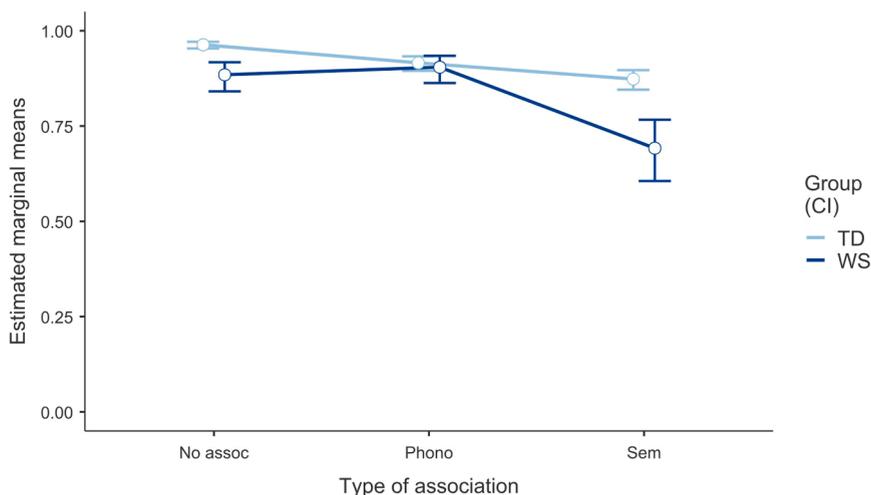
Fig. 2. *Estimated marginal means of answer accuracy by association type and group for the GLMM with EVIP as a covariate. Note.* 95 % CIs are presented.

Table 4
Summary table of estimated accuracies and associated statistics for the GLMM including the RVPM-2 as a covariate.

group	link	Acc.	SE_{acc}	group	Link	Acc.	SE_{acc}	exp(B)	$SE_{exp(B)}$	$p_{bonferroni}$
WS Group performance comparison										
WS	no assoc	0.899	0.019	WS	phono	0.916	0.017	0.811	0.114	1.000
WS	no assoc	0.899	0.019	WS	sem	0.724	0.043	3.384	0.412	< .001
WS	phono	0.916	0.017	WS	sem	0.724	0.043	4.171	0.623	< .001
TD Group performance comparison										
TD	no assoc	0.962	0.005	TD	phono	0.913	0.010	2.415	0.272	< .001
TD	no assoc	0.962	0.005	TD	sem	0.869	0.014	3.820	0.404	< .001
TD	phono	0.913	0.010	TD	sem	0.869	0.014	1.582	0.168	< .001
WS vs TD performance										
WS	no assoc	0.899	0.019	TD	no assoc	0.962	0.005	2.856	0.722	< .001
WS	phono	0.916	0.017	TD	phono	0.913	0.010	0.960	0.257	1.000
WS	sem	0.724	0.043	TD	sem	0.869	0.014	2.530	0.642	.004

Note. Link: type of association (no assoc = no association, phono = phonological, sem = semantic); *Acc.*: estimated accuracy; SE_{acc}: standard error of accuracy; exp *(B)*: odds ratio; $SE_{exp\ (B)}$: standard error of odds ratio; $p_{bonferroni}$: Bonferroni-adjusted p-value.

Table 5
Summary of scores to the EVIP and RVM-2 tests.

Tests	Group	N	Mean	Median	SD	Min	Max	test	stat	p
EVIP raw score	WS	19	81.3	90	34.2	16	143	U	168	.715
	TD	19	82.6	88	17.3	46	105			
RVPM−2 aptitude score	WS	19	426.4	433	30.3	363	469	t	1.665	.105
	TD	19	441.8	433	26.9	399	505			

Note. For EVIP, Levene test was significant, so we used Mann-Whitney U.

4. Discussion

The aim of this study was to examine the sensitivity to phonological and semantic information in WS, in order to re-examine the phonological-semantic imbalance hypothesis of language processing, by furthermore using a within-participant, single-task design and recruiting a homogeneous group of WS children in terms of chronological age.

The different analyses, involving either an estimate of verbal or non-verbal intellectual efficiency as covariates, led to convergent results. Children with WS performed similarly to TD children in the phonological condition but significantly worse in the semantic condition of the word association judgment task. Although TD children also showed slightly lower performance in the semantic condition compared to the phonological one, the performance gap was much larger in the WS group. These results appear to be consistent with the overall phonological-semantic imbalance hypothesis and provide some answers to three sub-hypotheses formulated in this context. More specifically, with regard to the atypical phonological representation hypothesis (sub-hypothesis 2), we observed that WS children showed developmental age-appropriate sensitivity to phonological information contradicting the idea of

Table 6

Summary table of estimated accuracies and associated statistics for the GLMM with the young TD group.

group	link	Acc.	SE$_{acc}$	group	Link	Acc.	SE$_{acc}$	exp(B)	SE$_{exp(B)}$	p$_{bonferroni}$
WS Group performance comparison										
WS	no assoc	0.825	0.031	WS	phono	0.854	0.029	0.811	0.114	1.000
WS	no assoc	0.825	0.031	WS	sem	0.583	0.054	3.385	0.413	< .001
WS	phono	0.854	0.029	WS	sem	0.583	0.054	4.172	0.624	< .001
WS Group performance comparison										
TD	no assoc	0.942	0.013	TD	phono	0.871	0.026	2.405	0.386	< .001
TD	no assoc	0.942	0.013	TD	sem	0.800	0.036	4.061	0.612	< .001
TD	phono	0.871	0.026	TD	sem	0.800	0.036	1.689	0.259	.009
TD vs WS performance										
WS	no assoc	0.825	0.031	TD	no assoc	0.942	0.013	3.443	1.091	.001
WS	phono	0.854	0.029	TD	phono	0.871	0.026	1.162	0.383	1.000
WS	sem	0.583	0.054	TD	sem	0.800	0.036	2.870	0.906	.013

Note. Link: type of association (no assoc = no association, phono = phonological, sem = semantic); *Acc.*: estimated accuracy; *SE$_{acc}$*: standard error of accuracy; exp *(B)*: odds ratio; *SE$_{exp\ (B)}$*: standard error of odds ratio; p$_{bonferroni}$: Bonferroni-adjusted p-value.

more variable phonological representations that would remain too sensitive to within-phoneme acoustic variations for the same phonemes occurring in different words or word contexts. Although some studies have reported less robust phonological processes (Pérez et al., 2022), our data suggest relative preservation, at least regarding incidental sensitivity to phonological associations between words. However, our protocol does not allow us to directly assess the impact of low-level acoustic processes on these representations, nor the hypothesis of excessive dependence on phonological working memory (sub-hypothesis 1; Brock, 2007; Grant et al., 1997; Thomas et al., 2001). Secondly, our results reveal a specific deficit regarding integrated processing of semantic information in WS children (sub-hypothesis 3), by showing significantly reduced performance for detecting semantic but not phonological overlap in two verbal stimuli. This deficit, which has already been documented (Jacobson & Smith Cairns, 2010; Laing, 2002; Temple, 2003), confirms that children with WS show difficulties in processing semantic-levels of information (Garayzábal Heinze & Cuetos Vega, 2010; Majerus et al., 2003; Romero-Rivas et al., 2025; Vicari et al., 1996). Finally, regarding sub-hypothesis 4, our results suggest that semantic processes are atypical rather than delayed given that the difficulties in detecting semantic associations are present even after controlling for overall verbal and nonverbal development, via the covariate or the matched-group statistical analyses. These results support the idea that phonological and semantic components may follow distinct pathways in WS children. Critically, the present study shows that this imbalance is not due to differences in phonological vs semantic task requirements as we assessed sensitivity to phonological and semantic information in an incidental manner within the same task context. Also, the results cannot be due to differences between WS group characteristics as a single group of WS participants completed both the phonological and semantic conditions.

Finally, we should note that the results observed in TD children slightly differed from those observed in a preceding study, which had aimed at validating the word association judgement task in TD children. In that study (Hippolyte et al., 2025), TD children identified semantic associations more effectively than phonological associations, in contrast to the present study. A likely explanation is the fact that in the validation study, the semantically related trials and the phonologically related trials were presented in separate task runs, and children needed to examine the word pairs only for one type of association at the same time. The present study shows that when TD participants are given the possibility to detect any type of association, they appear to privilege phonological associations. This observation raises interesting questions about attentional focus and task demands in linguistic processing, suggesting that when both types of associations must be monitored simultaneously, phonological similarities may be more salient or require fewer cognitive resources to detect than semantic relationships. This observation is also in line with similar results regarding differential sensitivity to phonological vs. semantic information in verbal working memory tasks (e.g., Kowialiewski et al., 2023).

4.1. Limitation of the study

The sample size was limited due to the practical constraints of being able to recruit a large sample of WS children. Given the rarity of this genetic condition, as well as the narrow recruitment age range of this study, our WS sample size remained limited. A power analysis using the simr package (Green & MacLeod, 2016; Kumle et al., 2021) indicated that a sample size of minimum 30 participants per group would be needed to achieve sufficient statistical power (e.g., >80 %) for a medium target effect size. Despite being lower than N = 30, we should note that the sample size of this study is considerably larger than that of many previous studies on the same population with the additional constraint of a restricted age range. We believe that the current sample represents the most comprehensive and balanced cohort feasible under the constraints of a rare condition, a small age range (Kover & Atwood, 2013), and real-world recruitment challenges. Furthermore, our design also included multiple observations per participant, allowing for both item-level and group-level analyses. This approach further enhances the robustness of our statistical inferences by making full use of the repeated-measures structure of the data (Toffalini et al., 2021).

One potential issue of this study is that we included EVIP and RVPM-2 scores as covariates to control for differences in receptive vocabulary and nonverbal reasoning between the WS and TD groups. Since the groups differed on these measures, adjusting for them could lead to ambiguous interpretation of the results (i.e. Lord, 1967). At the same time, we should note that the re-analysis without covariates on a younger TD control group matched on EVIP scores with the WS group had led to the same results as the analysis

including the covariates: the WS group still showed a disproportionately lower performance in the semantic condition compared to the phonological condition. This suggests that our main conclusion—that semantic processing in WS is atypical rather than delayed—is robust across different analytic approaches.

4.2. Conclusion

The results of this study provide robust novel evidence for the existence of a phonological-semantic imbalance during language processing in WS. They support the idea that the contrasted language profile documented in WS, characterized by fluent and proficient language production but often contextually inadequate verbal content, may be due to atypical semantic representations and/or processing. Our findings suggest that language intervention for individuals with WS should primarily focus on the development of semantic and conceptual processing. This targeted approach could include explicit teaching of semantic relationships, and conceptual networks, via targeted activities. While phonological processing may also show some specificities, it does not appear to have a major impact on receptive oral language processing, at least not in the context of the task used in this study. Future research should deepen these findings by examining the impact of the phonological-semantic imbalance on naturalistic language use and associated communicative difficulties in individuals with WS.

CRediT authorship contribution statement

Steve Majerus: Writing – review & editing, Validation, Supervision. **Amandine Hippolyte:** Writing – review & editing, Writing – original draft, Resources, Methodology, Investigation, Formal analysis, Data curation. **Laure Ibernon:** Writing – review & editing, Validation, Supervision, Project administration, Funding acquisition, Conceptualization. **Nathalie Marec-Breton:** Writing – review & editing, Conceptualization. **Christelle Declercq:** Writing – review & editing, Conceptualization.

Ethical considerations

ID-RCB: 2022-A00341–42

Declaration of Competing Interest

The authors declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Appendix A. Supporting information

Supplementary data associated with this article can be found in the online version at doi:10.1016/j.ridd.2025.105134.

Data Availability

I have shared the link to my data in the paper. Data are available at the following link: https://osf.io/ukg6b/?view_only=b54010ade17e482586edc9c142233f95.

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