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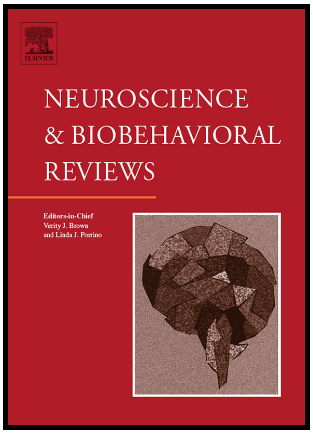

Acoustic and phonological processes in Williams Syndrome: a systematic review and meta-analysis

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Acoustic and phonological processes in Williams Syndrome: a systematic review and meta-analysis

Acoustic and phonological processes in Williams Syndrome: a systematic review and meta-analysis

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Conflict of Interest

None of the authors have any conflicts of interest.

Declarations of interest

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Abstract (max 250 words)

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This meta-analysis focuses on acoustic, phonetic and phonological aspects of language processing in Williams syndrome (WS), a rare neurodevelopmental genetic disorder. Based on N=51 papers, we aimed at identifying the status of these languages processes in WS relative to different types of control groups and we examined possible sources of variability of results through moderator analyses. At the acoustic level, evidence for hearing loss and impaired acoustic discrimination was observed. At the phonetic and sublexical phonological level, results were inconclusive due to limited literature. At the metaphonological level, WS individuals demonstrated better phonological awareness than nonverbal age-matched peers but performed below typically developing (TD) peers matched for verbal mental age. For phonological working memory WS performed worse than mental age matched TD peers but outperformed participants with other disabilities. At the lexical phonological level, WS demonstrated better phonological fluency skills than younger mental age matched peers. A wide range of heterogeneity was observed (I^2=0% to 92.26%). In some cases, the heterogeneity was partly explained by differences in control groups, but the largest part of heterogeneity could not be accounted for by the moderator variables included in the analysis. Future research needs to address sublexical phonological levels, to consider developmental trajectories and WS group variability, and to examine how impaired acoustic processes impact linguistic processes.

Keywords

Williams syndrome, meta-analysis, acoustic discrimination, categorical perception, phonotactic representations, phonological working memory, phonological fluency.

1. Introduction[2]

Williams syndrome (WS) is a rare genetic neurodevelopmental disorder with a prevalence varying from one birth in 7,500 to 1 in 20,000 (Morris et al., 1988; Strømme et al., 2002) and characterized by mild to moderate intellectual deficiency (ID) (Bellugi et al., 1994). The well-developed conversational abilities observed in individuals with WS were initially taken as

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[2] Abbreviations used:
WS: Williams syndrome; OD: other disabilities control group; MR: mixed etiology control group; CA: chronological age; MA: mental age; VMA: verbal mental age; NVMA: nonverbal mental age

evidence for the independence between language and other cognitive functions (Bellugi et al., 1993), but subsequent research revealed language difficulties (Laing et al., 2001; Rossi et al., 2012; Sampaio et al., 2008; Vicari et al., 2004; Volterra et al., 1996), supporting a neuroconstructivist approach instead (Thomas & Karmiloff-Smith, 2003). Thomas and Karmiloff-Smith furthermore proposed that WS may be characterized by an imbalance between phonological and semantic processes, with a stronger reliance on phonological levels of processing than semantic. Romero-Rivas et al. (2023) conducted a meta-analysis on lexical-semantic processes in WS. They showed inferior lexical-semantic abilities in WS compared to typically developing individuals but better abilities relative to other neurodevelopmental disorders. However, regarding phonological processes and associated acoustic processes, no meta-analysis has been conducted so far. Almost two decades ago, Brock (2007) conducted a review on various aspects of language abilities in WS, from phonology to pragmatic skills. However, the scarcity of reviewed studies prevented to conduct a meta-analysis per language component. To date, no meta-analysis has been conducted specifically on phonological processes and associated acoustic processes in WS, despite of the fact that these processes are fundamental building blocks of language development. This gap significantly limits our understanding of this rare syndrome and its neurodevelopmental specificities. We used a metanalytic approach to resolve inconsistencies between individual studies often limited by small sample sizes, to detect effects that might be masked in isolated studies, and to allow for a systematic evaluation of the influence of moderator variables such as control group type or experimental methodologies.

The aim of this study is to conduct an up-to-date meta-analysis of acoustic and phonological abilities in individuals with WS. For the most basic, acoustic levels of processing, we examined the literature on hearing acuity and thresholds (pure tone audiometry; transient evoked otoacoustic emission – TEOAE, distortion product otoacoustic emission - DPOAE) and acoustic discrimination (differentiation of two acoustic stimuli). At the segmental phonological level, we examined the literature on phonological discrimination abilities (judging whether two verbal sounds differ by a single phoneme or not) as well as categorical perception abilities, which involve detecting acoustic differences in language sounds stemming from the same or different phonemic categories (Liberman et al., 1967). At the suprasegmental phonological level, we focused on phonotactic levels of processing which involve knowledge about the co-occurrence and transitional probabilities of phoneme associations. This statistical knowledge about verbal sound association patterns allows for efficient segmentation of continuous speech and the identification of word boundaries (Saffran et al., 1996). At a metaphonological level, we examined the literature on phonological awareness. Phonological awareness is the ability to understand that words are made of individual sounds, which, furthermore, can be manipulated. Metaphonological awareness ranges from awareness to word parts (syllables, rimes, onsets) to isolated sounds (Treiman & Zukowski, 1991) and is an important predictor of reading acquisition. Developing phonological awareness greatly contributes to the advancement of oral and written language skills (Al Mannai & Everatt, 2005). An associated ability we reviewed is the maintenance of phonological information in working memory. This ability plays a critical role in phonological awareness but also in word and reading acquisition (Gathercole & Baddeley, 1989; Leclercq & Majerus, 2010; Martinez Perez et a., 2012; Ordonez Magro et al., 2020, 2021) as it enables individuals to hold novel phonological information in mind, to process it in a controlled manner and to repeat it. Finally, at the lexical phonological level, we focused on phonological fluency which reflects the capacity to generate words sharing a specific onset word form sound. It provides insight into the organization of the mental lexicon (Neergaard et al., 2019) although it should be noted that this task also has significant executive processing demands.

We conducted a multi-level meta-analysis for each of the specified acoustic and phonological processing levels, contingent upon sufficient data availability. A series of moderating variables were considered such as the type of task used to measure a specific acoustic/phonological level, the type of control group (typically developing participants, participants with Down syndrome (DS) or another population presenting with intellectual deficiency) and the article year of publication.

2. Methods

2.1. Meta-analysis

This meta-analysis was performed according to the Preferred Reporting Items for Systematic reviews and Meta-Analysis (PRISMA) guidelines (Page et al., 2021). This meta-analysis was not pre-registered.

2.2. Search strategy

A systematic literature search was performed on articles listed in PubMed, ScienceDirect, EBSCOhost (APA PsycArticles, APA PsycInfo and Psychology and Behavioral Sciences Collection) and Web Of Science, with no date restriction (first search conducted on June 28th, 2022, final search conducted on April 25th, 2025). Based on the different acoustic and phonological target processes defined above, the following search expression was used : ("pitch discrimination" OR "categorical perception" OR "acoustic" OR "phonological processing" OR "phonological awareness" OR "phonological discrimination" OR "phonotactics" OR "auditory processing" OR "phonological short term memory" OR "verbal short term memory" OR "auditory short term memory" OR "auditory working memory" OR "phonological fluency") and Williams Syndrome (such as "Williams syndrome" or “Williams Beuren syndrome”) (see Supplementary Material 1 for more precisions).

2.3. Selection of studies

Studies were screened and selected by AH and MP according to the following criteria. In order to be included, a study: 1) included a group of subjects with WS and compared them to existing norms or to a control group, 2) addressed at least one of the target processes defined above, 3) was peer-reviewed. Exclusion criteria were as follows 1) exclusion of the second of two studies that report experiments with the same WS population and identical tests, 2) studies reporting a composite score that combines several target acoustic/phonological processes, 3) studies lacking matching variables for WS and control groups. All studies were considered for meta-analysis except if the study did not provide sufficient numerical information for calculation of effect sizes or reported a case study.

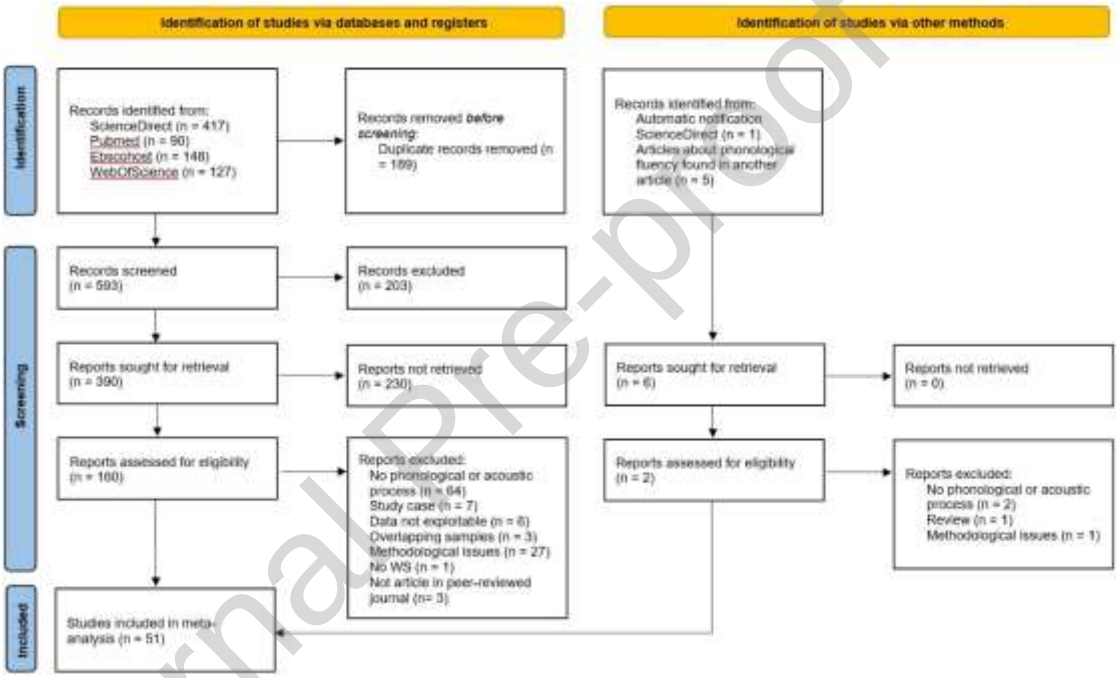

Fig. 1. Flow Diagram of the Systematic Review according to PRISMA (2020)

2.4. Data extraction

AH and MP extracted the following data: surname of the first author, year of publication, number of participants in WS and control groups, mean ages and age ranges, control group type, control group matching type, mean age of the sample, test used, significance of the results, means, standard deviations, interquartile range, median (See Supplementary Material 2 for more precisions).

Various tasks and measures were employed for each process, necessitating the computation of standardised mean differences (SMD). When means or standard deviations were not available, alternative summary statistics such as minimum, first quartile, median, third quartile, and maximum were utilized to estimate mean and SD, aligning with recommendations by Weir et al. (2018). In instances where numerical data were insufficient, values were estimated from graphical representations, as suggested by Koricheva, Gurevitch, and Mengersen (2013), by using the tool WebPlotDigitizer (https://apps.automeris.io/wpd4/). Graphical data extraction was independently performed by two scorers (AH and MP) on all relevant plots, in order to ensure reliability of the extracted data. Given the small sample sizes (N < 20) and notable differences in SD values between the WS and control groups, Hedges' g was employed as an effect size estimator for its capability to adjust for varying SDs. Calculation of Hedges' g followed formulas 1 and 4 outlined by Lakens (2013). When summary statistics were missing for SMD computation, effect size was derived from other statistical information (e.g., Cohen's d, ANOVA's F, Student's t, z from a statistical test, p value) using several Card's formula (2011). The R script is accessible at https://osf.io/9dprv/?view_only=168b8cd905b141df9d3e99ebef210a77. You can access interactive data visualization at https://sounds.shinyapps.io/data_visualization_meta_analysis_williams_syndrome/.[3]

2.5. Data analysis for the meta-analysis

Data analysis was conducted using the "Metafor" and "Dmetar" packages for R, version 4.3.2. (http://cran.r-project.org/). A three-level meta-analysis with a random-effects model using the restricted maximum likelihood (REML) estimator was fitted to calculate the effect size and the 95 % confidence interval (CI). Comparisons between mean effect sizes were made with the Knapp-Hartung method. The multi-level design allowed to control for dependencies between the effect sizes (such as the same WS group being compared to several control groups

[3] For readers who experience difficulty accessing the Shiny apps, the authors can provide the corresponding R code upon request. Alternatively, all code is openly available on the OSF repository.

or on different tasks). To assess small-study effects, we generated funnel plots and analysed them via the three-level version of Egger's regression test (Egger et al., 1997) developed by Rodgers and Pustejovsky (2021) (see Supplementary material 3 for the graphics). Influential data points were identified by using Cook's distance, which measures how much each observation affects the estimates of the regression model parameters, helping to pinpoint potential outliers or influential data points in the analysis. To assess between-study heterogeneity, we used absolute (Cochran's Q) and relative (I^2) measures. For I^2, we categorized values of 25%, 50%, and 75% as indicating low, moderate, and high heterogeneity, respectively. We evaluated heterogeneity across three levels: Level 1 of I^2 assessed the sampling variation of effect sizes (which is not included in the calculation of the overall I^2); Level 2 assessed the within-study variation of effect sizes, and Level 3 assessed the between-study variation of effect sizes. The overall I^2, termed I^2 general, measures the percentage of variability not attributable to sampling error and is composed of Levels 2 and 3. We furthermore calculated prediction intervals (PI, IntHout et al., 2016). In random-effect analyses, compiled studies are considered not to estimate the same true effect but to be heterogenous and to estimate various true effects. The PIs represent the range in which, based on the data obtained, we expect the true effect sizes to fall 95% of the times.

Moderator analysis was conducted to locate the source of heterogeneity in results. As mentioned above, three moderators were defined: type of task used, year of publication and type of control group. The Type of task moderator was redefined for each target process, as a function of the tasks used for assessing each target process. For acoustic discrimination, the Type of task moderator modalities were 'sound' (e.g., tone, chords, melodies) and 'rhythm' (rhythmic pattern). For phonological awareness, we identified three Type of task moderator modalities based on the hierarchical view of the syllable model (Treiman & Zukowski, 1991): 'syllable awareness', 'onset and rhyme awareness' and 'phoneme awareness'. For phonological working memory, the Type of task moderator modalities were: 'sentence repetition', 'word repetition' (forward digit span, word span, etc.), 'nonword repetition' and 'central executive' (backward digit span, manipulation of information, score combining forward and backward digit span). Publication year was incorporated as a continuous moderator variable. Finally, the Control group moderator was defined according to the following modalities: typical development participants (TD), participants with other disabilities (OD) (i.e. Down syndrome, individuals with intellectual disability disorder of mixed etiology (MR), and children with moderate learning difficulties (MLD) caused by neurodevelopmental disorders or early life neurological lesions). These Control group modalities were further subcategorized based on the matching variables that were used (verbal tests → 'verbal mental age'; non-verbal tests → 'nonverbal mental age'; IQ test or other verbal-nonverbal combined test → 'mental age'; chronological age). If the groups were matched on both chronological age and mental age, we subcategorized as 'mental age'. We focused on these specific control group specifications as they are the type of control groups most typically used when assessing cognitive and linguistic abilities in WS. More specifically, comparison to a control group matched on chronological age allows for the identification of overall developmental lag relative to typical development. Comparison to a control group matched on verbal mental age allows for the identification of specific strengths and weaknesses in linguistic profiles relative to overall verbal abilities. Comparison to a control group matched on nonverbal mental age allows to situate the level of language development relative to nonverbal cognitive development. Finally, comparison to a control group matched on mental age allows to determine linguistic strengths and weaknesses relative to overall cognitive development.

3. Results

3.1. Auditory perception

3.1.1. Acoustic performance

Hedges' g couldn't be calculated due to significant variation of type of data presented across included studies for acoustic performance. We focused on prevalence rates, which were the most common type of data.

A total of k = 7 studies was included for assessing hearing loss, amounting to a sample size of n = 170 individuals with WS. The pooled prevalence of hearing loss was estimated to be 43.14%, 95% CI [29.26% - 58.18%] and was associated with substantial heterogeneity between studies (I^2 = 59.00%, Q(6) = 15.94, p = .0141). Regarding acoustic reflexes, a pooled prevalence could not be calculated due to highly diverse measurement methodologies. Regarding TEOAEs, a total of k = 3 studies was included, for a sample size of n = 49 individuals. The pooled prevalence of absent TEOAEs (indicating hearing loss) was estimated to be 63.27%, 95% CI[32.49%; 86.04%] with no heterogeneity (I^2 = 0.00%, Q(2) = 1.50, p = .4713). Finally, only one study explored DPOAEs, hence no further meta-analytic analyses were performed.

3.1.2. Acoustic discrimination

A total of k = 4 studies was retrieved with exploitable data, for a total of 64 individuals with WS (Fig. 2). In this process, the control groups consisted solely of TD participants, and these groups were matched on verbal mental age, mental age or chronological age. A large effect was observed, meaning that the WS groups showed decreased performance in acoustic discrimination relative to TD control groups (g = -1.3964, p = .0361). A high level of heterogeneity was identified (I^2 = 92.26%, Q(7) = 47.6463, p < .0001). The I^2_{Level2} amounted to 92.26% and the I^2_{Level1} amounted to 7.74, meaning that heterogeneity is mostly due to within-study heterogeneity and sampling variance. The hypothesis of a symmetrical funnel plot could not be rejected, although note that the p-value approached significance (intercept = 8.3791, 95% CI[-57.8396 ; 1.5407], t(6) = -2.3199, p = .0595). Year of publication was not a significant moderator (F(1;6) = 0.3827, p = .5589). Moderator analysis for control group and type of task could not be conducted due to the low number of studies for some of the modalities (i.e. < 2).

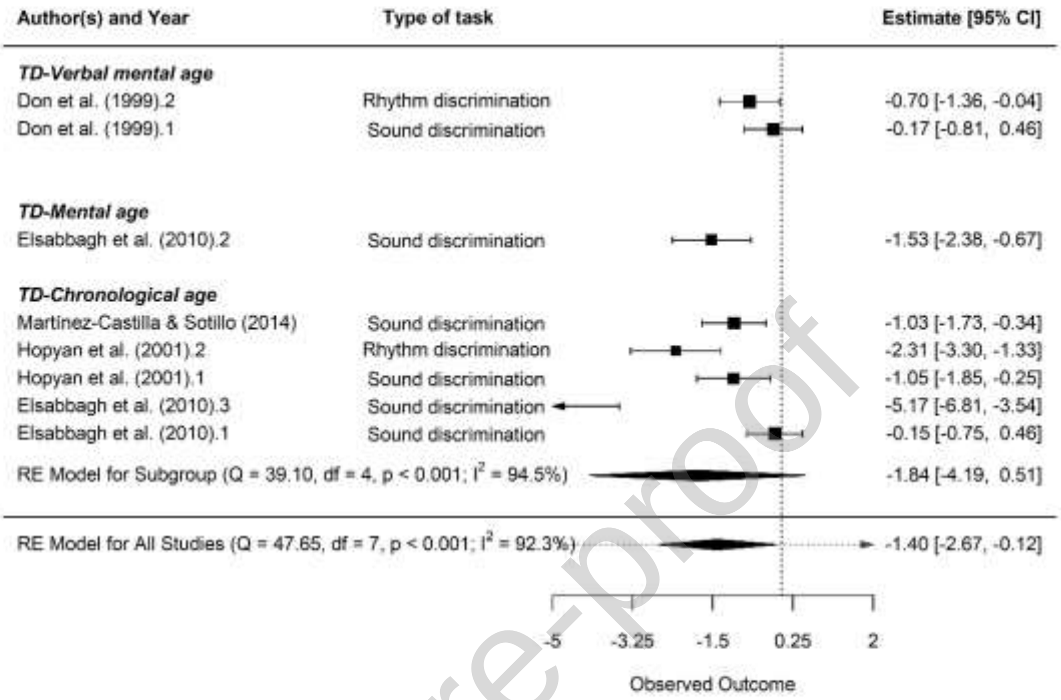

Fig. 2. Forest Plot for Acoustic Discrimination Based on Random Effect Model. Note: Q = heterogeneity test, df = degrees of freedom, p = p-value of the test, I^2 = percentage of heterogeneity, Q_M = moderator test, TD = typical development group control. The general effect size and 95% CI were omitted for TD-VMA and TD-MA because of too few effect sizes included in each modality (i.e. < 2).

3.1.3. Phonological discrimination

A total of k = 2 studies was retrieved with exploitable data for a total of 24 individuals with WS (Fig. 3). In this process, the control groups consisted solely of TD participants, and these groups were matched on verbal mental age or chronological age. A large negative effect was observed, meaning that the WS individuals performed significantly lower than the TD control group (g = -0.8912, p = .0006) in phonological discrimination. A low level of heterogeneity was identified (I^2 = 0.00 %, Q(2) = .0144, p = .9928). Due to the low number of studies, no further moderator analyses were performed.

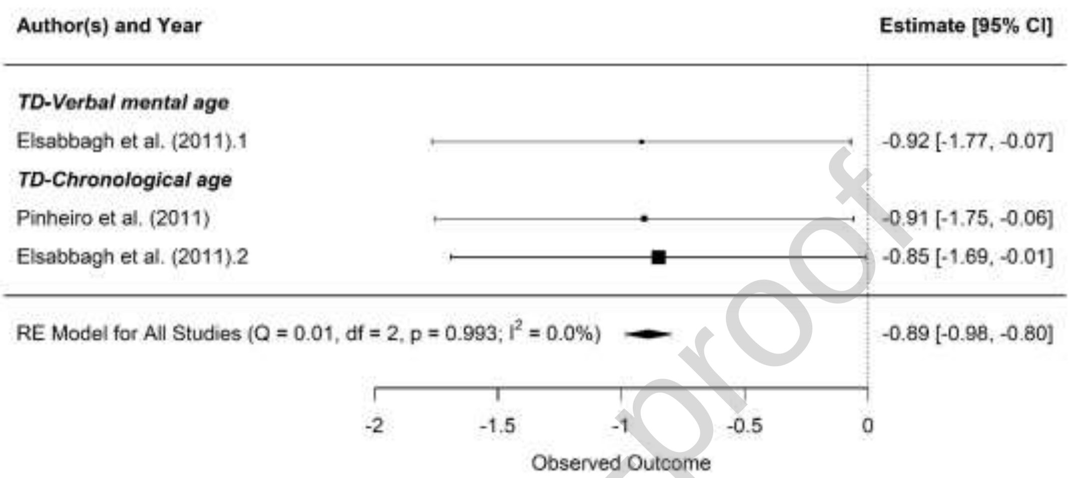

Fig. 3. Forest Plot for Phonological Discrimination Based on Random Effect Model. Note: Q = heterogeneity test, df = degrees of freedom, p = p-value of the test, I^2 = percentage of heterogeneity, TD = typical development group control.

3.1.4. Categorical perception

No studies meeting the predefined inclusion criteria were identified for the evaluation of categorical perception in individuals with WS.

3.2. Phonotactic representations

No studies meeting the predefined inclusion criteria were identified for the evaluation of phonotactic representations in individuals with WS.

3.3. Phonological awareness

A total of k = 10 studies was retrieved with exploitable data, for a total of 209 individuals with WS (Fig. 4). In this process, the control groups consisted of TD and DS participants. TD groups were matched on verbal mental age, mental age, nonverbal mental age or chronological age, while DS groups were only matched on mental age. Individuals with WS appeared to perform comparably to the combined performance of all control groups (g = -0.1529, p = 0.5433) in phonological awareness. A high level of heterogeneity was identified (I^2 = 89.87 %, Q(29) = 236.2950, p < .0001). I^2_{Level2} amounted to 44.75% and I^2_{Level3} amounted to 45.12%, indicating that total variance was evenly explained by within-study and between-study heterogeneity. A symmetrical funnel plot was observed (intercept = 0.6801, 95% CI[-9.6754 ; 4.1369], t(28) = -0.8214, p = .4184), indicating no evidence of publication bias.

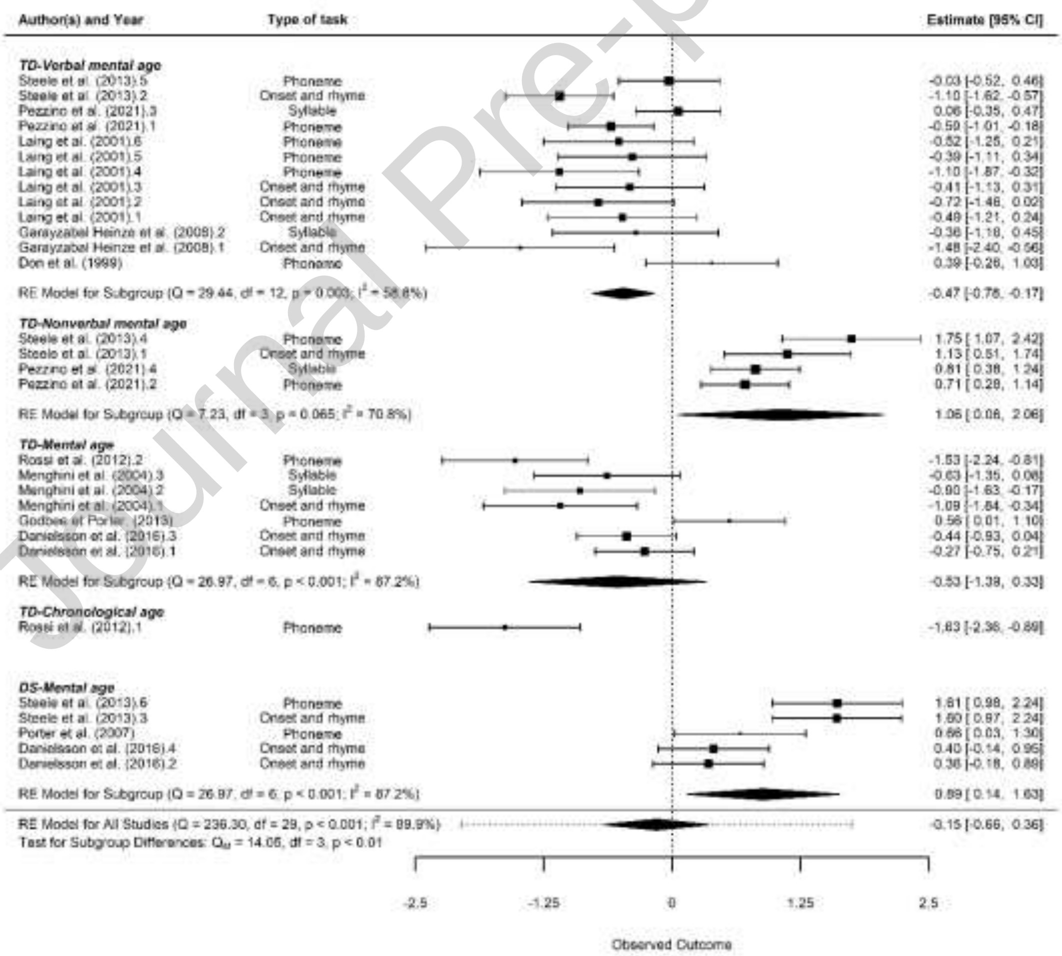

Fig. 4. Forest Plot for Phonological Awareness Based on Random Effect Model. Note: Q = heterogeneity test, df = degrees of freedom, p = p-value of the test, I^2 = percentage of heterogeneity, Q_M = moderator test, TD = typical development group control, DS = Down syndrome group control. The general effect size and 95% CI were omitted for TD-CA because of too few studies included in this modality (i.e. < 2).

The meta-regression using control group as a moderator showed a significant effect, $F(3, 25) = 14.0453$, $p < .0001$) . For this analysis, the fifth modality (i.e. TD-CA) was removed because it was only composed of one effect size. I^2 was reduced ($I^2 = 78.24\%$) but heterogeneity remained significant ($QE(25) = 80.8548$, $p < .0001$). Subgroup analyses (table 1) showed a small-to-medium negative effect when comparing WS vs. TD-VMA (n = 13), g = -0.4750, p < .001; no significant difference when comparing WS vs. TD-MA (n = 7), g = -0.5307, p = .1814; a large positive effect when comparing WS vs. TD-NVMA (n = 4), g = 1.0610, p = .0435; and a large positive effect when comparing WS vs. DS-MA (n = 5), g = 0.8864, p = .0295. See table 2 for details.

Table 1 and 2: phonological awareness subgroup analysis and comparisons

Type of task was not a significant moderator ($F(2, 27) = 0.3972$, $p = .6761$), neither was there a significant moderator effect of year of publication ($F(1, 28) = 0.7623$, $p = .3900$).

3.4. Phonological working memory

A total of k = 34 studies was retrieved with exploitable data, for a total of 700 individuals with WS (Fig. 5). In this process, the control groups consisted of TD and other disability groups (OD, Down syndrome participants, ID of mixed etiology, and moderate learning difficulties). TD groups were matched on verbal mental age, mental age, nonverbal mental age or chronological age, and OD groups were matched on mental age, nonverbal mental age, verbal mental age or chronological age. For phonological working memory, a small and non-significant negative effect size was observed ($g = -0.2142$, $p = .1694$), independently of moderators. A high level of heterogeneity was identified ($I^2 = 89.34$, $Q(83) = 620.1557$, $p < .0001$). 35.28% of variance was due to within-study heterogeneity (I^2_{Level2}) and 54.06% of variance was due to between-study heterogeneity (I^2_{Level3}). The funnel plot was symmetrical, suggesting no publication bias (intercept = -0.5295, 95% CI [-2.856 ; 4.7611], $t(82) = 0.4975$, $p = .6201$).

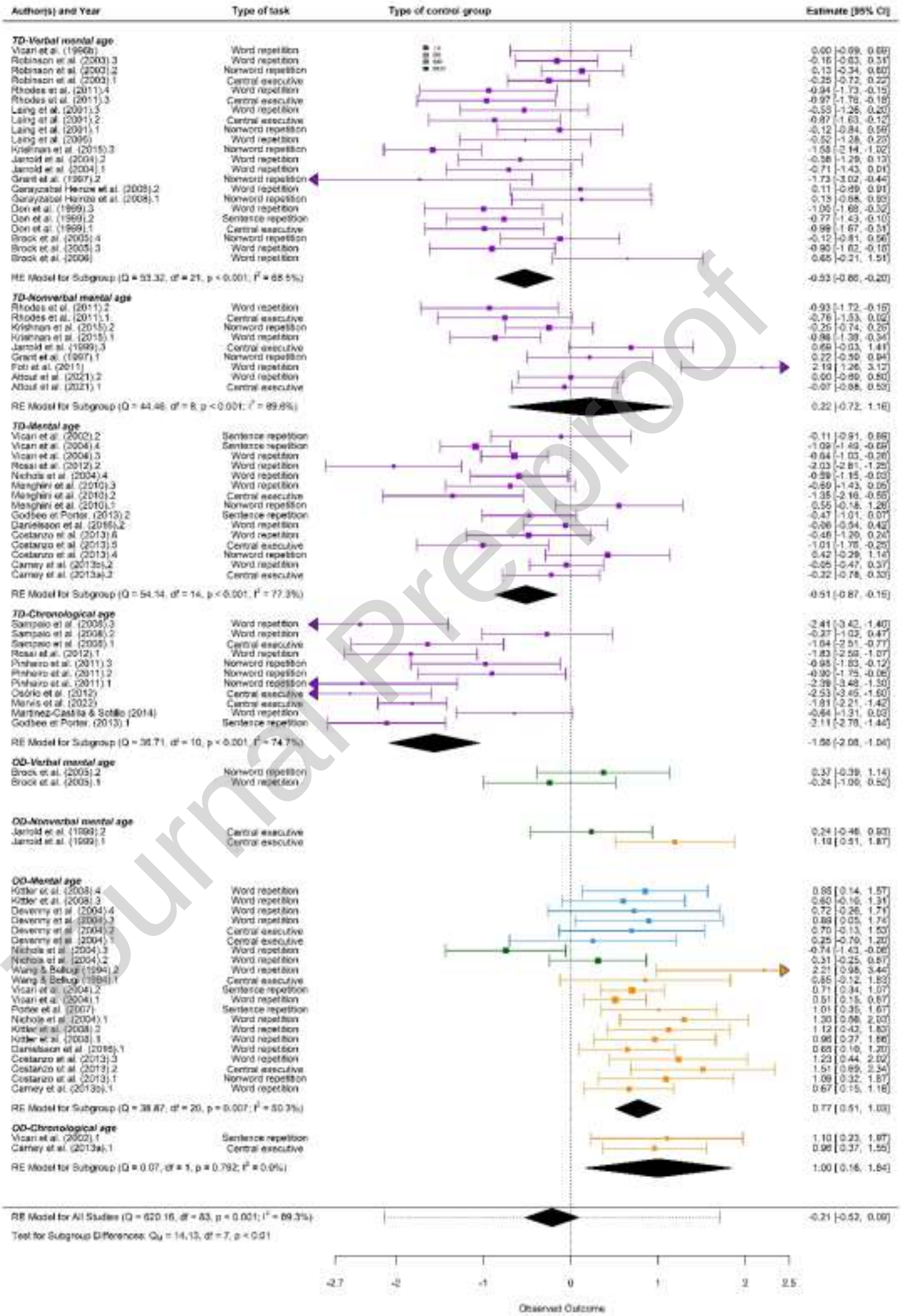
Author(s) and Year
Type of task
Type of control group
Estimate [95% CI]
TD-Verbal mental age
TD-Nonverbal mental age
TD-Mental age
TD-Chronological age
OD-Verbal mental age
OD-Nonverbal mental age
OD-Mental age
OD-Chronological age
Test for Subgroup Differences: Q_M = 14.13, df = 7, p < 0.01
-2.7
-2
-1
0
1
2
2.5
Observed Outcome

Fig. 5. Forest Plot for Phonological Working Memory Based on Random Effect. Note: Q = heterogeneity test, df = degrees of freedom, p = p-value of the test, I^2 = percentage of heterogeneity, Q_M = moderator test, TD = typical development group control, OD = Other disabilities, DS = Down syndrome group control, MR = mixed etiology control group, MLD = moderate learning difficulties control group. The general effect size and 95% CI were omitted for OD-VMA, OD-NVMA and OD-CA because of too few studies included in each modality (i.e. < 2).

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The meta-regression using control group as a moderator showed an effect, $F(4, 73) = 21.7941$, $p < .0001$). I^2 was reduced ($I^2 = 71.5\%$) but heterogeneity remained significant ($QE(73) = 227.5047$, $p < .0001$). Subgroup analyses (table 3) showed a large negative effect when comparing WS vs. TD-CA (n = 11), g = -1.5609, p < .0001; no difference when comparing WS vs. TD-NVMA (n = 8), g = 0.2242, p = 0.5971; a medium negative effect when comparing WS vs. TD-VMA (n = 22), g = -0.5301, p = .0033; a medium negative effect when comparing WS vs. TD-MA (n = 15), g = -0.5105, p = .0085; and a medium-to-large positive effect when comparing WS vs. OD-MA (n = 21), g = 0.7738, p < .0001. The other comparisons are not presented due to insufficient studies retrieved in each modality. See table 4 for details.

Table 3 and 4: Phonological working memory subgroup analysis and comparisons

Type of task showed no significant moderating effect ($F(3, 79) = 0.9260$, $p = 0.4322$) and did not reduce heterogeneity ($I^2 = 89.09\%$). The same was true for year of publication, $F(1, 82) = 3.2669$, $p = .0744$.

3.5. Phonological fluency

A total of k = 4 studies was retrieved with exploitable data, for a total of 111 individuals with WS. In this process, the control groups consisted of TD and participants with OD, only matched on mental age. A positive effect was found (g = 0.6115, p = 0.0062) meaning that WS performed better than mental age-matched control groups on phonological fluency tests. Heterogeneity was low and non-significant (I^2 = 8.78 %, Q(6) = 10.5098, p = .1048). I^2_{Level2} was equal to I^2_{Level1} (8.78%), meaning that heterogeneity was exclusively due to within-study heterogeneity. The funnel plot was symmetrical (intercept = 0.8080, 95% CI[-3.6398; 2.0345], t(5) = -0.7273, p = .4997), suggesting no publication bias. Year of publication (F(1;5) = 0.1045, p = .7596) was not a moderator. Control group moderator analysis could not be made due to the low number of studies for the modality OD-MA (i.e. < 2).

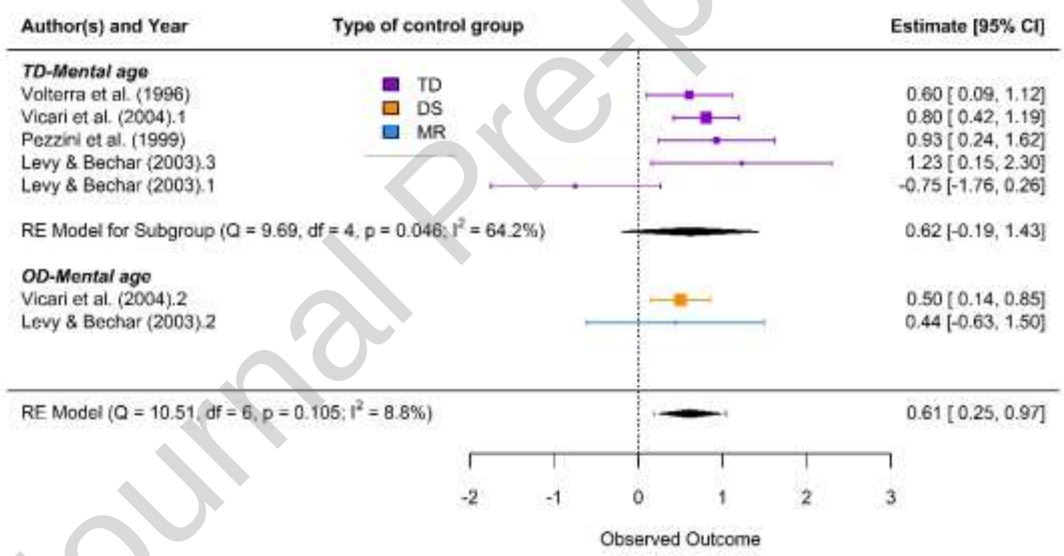

Fig. 6. Forest Plot for Phonological Fluency Based on Random Effect Model. Note: Q = heterogeneity test, df = degrees of freedom, p = p-value of the test, I^2 = percentage of heterogeneity, TD = typical development group control, OD = other disabilities, DS = Down syndrome group control, MR = mixed etiology control group.

Color must be used for the fig. 6.

4. Discussion

This meta-analysis examined acoustic and phonological abilities in individuals with WS, by focusing on eight different subdomains (hearing, acoustic discrimination, phonological discrimination, categorical perception, phonotactics representation, phonological awareness, phonological working memory and phonological fluency). Overall, a large heterogeneity was observed for most subdomains, with type of control group being a major moderator.

More specifically, regarding hearing abilities, studies using pure tone audiometry evidenced a high and variable prevalence of hearing loss. A high prevalence of absent TEOAEs was also noted. Other aspects could not be entered in the meta-analysis due to very low number of studies (for DPOAEs; see Marler et al., 2010) and the use of highly divergent measurement techniques (for acoustic reflexes; see Attias et al.,2008; Barrozi et al., 2012; Johnson et al., 2001; Zarchi et al., 2011). Overall, the meta-analytic finding of a high prevalence of hearing impairment based on pure tone audiometry and TEOAEs is in line with the assumption of atypical auditory processing caused by the 7q11.2 microdeletion and resulting elastin deficiency (Dridi et al., 1999). At the same time, prevalence rates showed a large variability and future studies need to determine the factors that may account for this heterogeneity.

Regarding acoustic and phonological discrimination abilities, this meta-analysis showed poorer acoustic and phonological discrimination performance in individuals with WS when compared to TD participants. On the one hand, these results may be expected given the high prevalence of hearing loss in WS. On the other hand, these results may appear surprising given that individuals with WS are often reported to show a strong attraction to auditory stimuli, including music (Levitin et al., 2004); (Thakur et al., 2018). One aspect that needs to be considered here is the 'same/different' response mode that is used in most studies. The concepts of 'identical' and 'different' may not always be easy to grasp for individuals with intellectual deficiency such as individuals with WS. Attentional and working memory difficulties could also lead to reduced performance on discrimination tasks (see below). At the same time, attraction to auditory stimuli and impaired auditory processing abilities are not necessarily mutually exclusive dimensions.

Regarding phonological awareness, a nuanced pattern of results was observed. While WS individuals outperformed TD-NVMA (i.e. TD peers matched on nonverbal mental age) and DS individuals, indicating a relative strength, they lagged behind TD-VMA individuals (i.e. TD peers matched on verbal mental age). The fact that WS individuals showed poorer abilities in phonological abilities only relative to language-matched peers could partially stem from the verbal matching variable that was used. This variable was most often a vocabulary task. Vocabulary knowledge has indeed been shown to be a strength in WS relative to other language domains (Romero-Rivas et al., 2023). At the same time, given that the I^2 was only marginally reduced when taking into account the 'type of group' moderator, other factors accounting for heterogeneity need to be considered such as the diversity of the cognitive profiles that characterize WS. For example, Porter and Coltheart (2005) highlighted the extreme variability of individuals with WS on phonemic awareness tasks, with individual z-scores for performance on this tasks ranging from -18 SD to +20 SD relative to an estimate of their own cognitive ability based on 21 different tests.

Regarding phonological working memory, our meta-analysis revealed a similar, nuanced pattern of results as for phonological awareness, with WS individuals again outperforming individuals with other types of intellectual deficiency but lagging behind individuals with TD-VMA. The matching on vocabulary knowledge may again explain this situation, at least in part. Overall, the relative strength in phonological working memory for individuals with WS as revealed in this meta-analysis is in line with early views of the cognitive profile of WS (e.g., Bellugi et al., 1993). It should however also be noted that this strength in phonological WM may depend upon the type of verbal stimuli being used (words vs. wordlike nonwords vs. nonwordlike nonword) (Majerus et al., 2003; Vicari, 1996), even if the present meta-analysis was not able to formally examine this question due to a low number of relevant studies.

With respect to phonological fluency in WS, our findings indicate performance that does not differ from control groups. These results can be considered to be robust as they were characterized by highly reduced heterogeneity. These results are also corroborated by two additional studies that were not included in this meta-analysis due to the specific reporting methods that were used (i.e. group mean percentile, Menghini et al., 2011, 2013). It should however also be noted that we were not able to perform a moderator analysis due to the limited number of relevant studies. More generally, these results support the hypothesis of imbalanced phonological and semantic processing abilities (Thomas & Karmiloff-Smith, 2003), by showing relatively preserved phonological fluency while a meta-analysis on semantic fluency showed impaired abilities for the latter (Romero-Rivas et al., 2023).

Regarding the categorical perception and phonotactic processing domains, we were not able to use a meta-analytic approach due to a very low number of studies having investigated these domains. We should briefly note here two multiple case studies that examined these domains. Majerus et al. (2011) assessed categorical perception for /d/-/t/ and /b/-/d/ consonant continua in six individuals with WS. They observed diminished between-category discrimination and enhanced within-category discrimination peaks in four out of six individuals with WS, with relatively inconsistent and heterogeneous patterns among individuals. Majerus et al. (2003) examined sensitivity to phonotactic processing by presenting nonword repetition tasks to four WS children, the nonwords being composed of frequent or infrequent phonotactic patterns relative to the children's native language. An atypical phonotactic frequency effect was observed, with age appropriate performance for low frequency but not high frequency nonwords. These two studies indicate the possible existence of atypical sublexical phonological processing abilities in WS, in line with the reduced hearing and phonological discrimination results mentioned earlier. It has been hypothesized that these specificities may represent one of the factors explaining poor phonological awareness and reading acquisition in WS (Majerus et al., 2003). Indeed, categorical perception is foundational for accurate and fast speech sound identification (Liberman et al., 1957), while phonotactic knowledge supports word segmentation (Saffran et al., 1996). This meta-analysis shows that a more systematic assessment of these findings with larger group studies remains necessary to confirm this hypothesis.

At a broader level, while this meta-analysis shows relative strengths in a number of phonological domains, it does not support a complete preservation of these abilities as individuals with WS nevertheless performed lower than TD individuals. This challenges the modularity approach (Fodor, 1986) which considered WS as genetic test case of the modularity of language relative to other cognitive domains. The nuanced pattern of results we observed, with a number of relative strengths (e.g., phonological fluency) but also with a number of areas with clear weaknesses (e.g., hearing abilities, acoustic and phonological discrimination), is more in line with a neuroconstructivist approach (Thomas & Karmiloff-Smith, 2003), considering that the cognitive profile of WS is atypical, with both strengths and weaknesses in different cognitive subdomains, including language.

4.1. Limitations and future studies

Due to the data aggregation approach that is inherent to a meta-analytic approach and due to the relative low number of studies, differences in cognitive profiles between individuals with WS and associated moderators could not be examined in a robust manner. Given the importance of interindividual differences that have been highlighted in WS (Miezah et al., 2020), the results of the present meta-analysis need to be considered with caution, particularly if the aim is to make predictions regarding phonological abilities in a specific person presenting with WS. These differences in cognitive profile, as well as differences in inclusion criteria, age ranges, and methodologies designs across studies, may be responsible for the heterogeneity that remains unexplained. These disparities limit comparability of the studies. Future studies may adopt common research protocols that would facilitate data sharing and accumulation across research centres, ultimately leading to larger data sets with increased statistical power. Without such coordination, it remains difficult to interpret between-study variability or fully explain residual heterogeneity in studies involving a rare disorder such as WS.

A further caveat is also needed regarding the type of control groups that were most frequently used in the studies included in this meta-analysis. The control groups were mainly TD individuals or individuals with DS. Given that DS is characterized by important language difficulties (Abbeduto et al., 2007), the relative phonological strengths observed in this meta-analysis for people with WS need to be considered with caution. Future studies should compare participants with WS with participants showing other types of neurodevelopmental disabilities, with and without ID, in order to allow for a more comprehensive assessment and understanding of the relative strengths and weaknesses of language abilities in WS.

Another limitation is the lack of control of chronological age - we did not distinguish between children, adolescents and adults with WS in the different analyses. Due to the rarity of the WS syndrome, most studies include participants from different age groups in order to optimize statistical power, with often very large age ranges. Future studies may adopt single-case meta-analytic designs by running meta-analysis over individuals rather than over group results, which would allow taking into account age moderators even for such rare syndromes as WS (see Maric et al., 2023, for an example on single case behavioural interventions).

4.2. Conclusion

The present study supports the conclusions from an earlier review paper (Brock; 2007) by showing that people with WS show decreased performance in a set of phonological processing tasks relative to TD or verbal mental age matched controls, but a relative preservation relative to participants with other disabilities. Hearing loss and impaired acoustic discrimination in WS was an overall consistent finding. Although phonological awareness appeared to be relatively unaffected - with individuals with WS obtaining better results than peers matched on non-verbal age and those with DS - it remained below chronological-aged appropriate levels. Conversely, phonological working memory abilities were weaker relative to TD children matched on mental age, but higher relative to other groups with a disability. Finally, phonological fluency skills appeared to reach high levels of development, people with WS outperforming younger TD children matched on mental age. Altogether, the variability between sub-domains suggests atypical developmental trajectories, supporting a neuroconstructivist approach to development in WS. To allow for a more complete understanding of acoustic and phonological processing profiles in WS, more extended research is needed regarding sublexical phonological processing, and more specifically categorical perception and phonotactic levels of processing. At the same time, this meta-analysis revealed a high degree of heterogeneity, that could not be fully accounted for by the moderator analyses. Given the rarity of WS, future studies may use single-case meta-analytic designs to allow for a more complete understanding of auditory and phonological processing abilities in WS.

Glossary

Acoustic Discrimination: Differentiation of two acoustic stimuli.

Phonological Discrimination: Judging whether two verbal sounds differ by a single phoneme or not.

Categorical Perception: Detecting acoustic differences in language sounds stemming from the same or different phonemic categories.

Phonotactic Representations: Knowledge about the co-occurrence and transitional probabilities of phoneme associations.

Phonological Awareness: Ability to understand that words are made of individual sounds which can be manipulated.

Phonological Working Memory: Enables individuals to hold novel phonological information in mind, process it in a controlled manner, and repeat it.

Phonological Fluency: Capacity to generate words sharing a specific onset word form sound.

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All articles included in statistical analyses have an asterisk at the start.

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Tables:

Table 1

Subgroup analysis for phonological awareness

Control group	Number of effect sizes	I²	g (general effect size)	95%CI	p-value
TD-NVMA	4	70.77	1.0610	0.0579 – 2.0641	0.0435*
TD-VMA	13	58.79	-0.4750	-0.7756 – -0.1743	0.0049**
TD-MA	7	87.18	-0.5307	0.1814 – -1.3898	0.1814
DS-MA	5	80.46	0. 8864	0.1439 – 1.6288	0.0295*

Note. * : p < .05, ** : p < .01. I² = percentage of heterogeneity, 95%CI = 95% Confidence Interval of the effect size, p-value = p-value of the test, TD = typical development group control, DS = Down syndrome group control, NVMA = nonverbal mental age, MA = mental age, VMA = verbal mental age.

Table 2

Between-subgroups differences analysis for phonological awareness

	TD-NVMA		TD-MA		TD-VMA		DS-MA	
	g	p	g	p	g	p	g	p
TD-CA	2.2791	-0.0028**	0.7122	-0.2573	0.8251	-0.2109	1.9226	.0066**
TD-NVMA			-1.567	0.001***	-1.4541	<.0001***	0.0432	.9023
TD-MA					0.1129	-0.7514	1.1975	.0009***
TD-VMA							1.4362	.8013***

Note. * : p < .05, ** : p < .01, *** : p < .001. g = effect size, p = p-value of the test, TD = typical development group control, DS = Down syndrome group control, NVMA = nonverbal mental age, MA = mental age, VMA = verbal mental age.

Table 3

Subgroup analysis for phonological working memory

Control group	Number of effect sizes	I^2	g (general effect size)	95%CI	p-value
TD-CA	11	74.71	-1.5609	-2.0781 – -1.0438	< .0001***
TD-NVMA	8	89.59	0.2242	-0.7152 – -1.1635	.5971
TD-VMA	22	68.5	-0.5301	-0.8625 – -0.1977	.0033**
TD-MA	15	77.27	-0.5105	-0.8684 – -0.1526	.0085**
OD-MA	21	50.29	0.7738	0.5132 – 1.0344	< .0001***

Note. * : p < .05, ** : p < .01, *** : p < .001. I^2 = percentage of heterogeneity, 95%CI = 95% Confidence Interval of the effect size, p-value = p-value of the test, TD = typical development group control, OD = other disabilities, CA = chronological age, NVMA = nonverbal mental age, MA = mental age, VMA = verbal mental age.

Table 4

Between-subgroups differences analysis for phonological working memory

	TD-NVMA		TD-MA		TD-VMA	
	g	p	g	P	g	P
TD-CA	1.7162	< .0001***	1.0506	.0008***	1.0716	.0006***
TD-NVMA			-0.6655	.0367*	-0.6445	.0235*
TD-MA					0.021	.9355

Note. * : p < .05, ** : p < .01, *** : p < .001. g = effect size, p = p-value of the test, TD = typical development group control, NVMA = nonverbal mental age, MA = mental age, VMA = verbal mental age.

Highlights

- WS participants show impaired acoustic discrimination.
- WS participants show reduced phonological awareness/working memory

- WS participants show a relative strength in phonological fluency
- Results are highly heterogeneous and depend on the type of control group
- Future studies should explore categorical perception and phonotactic representations