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Cognitive Profiles of French Individuals with Williams Syndrome and Down Syndrome: What's Up? What's Next?

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Abstract: The aim of our study was to characterize the face recognition skills, theory of mind skills, and language skills of a single group of French participants with Williams syndrome (WS) compared to a group of participants with Down syndrome (DS). Twelve French-speaking participants with WS and 12 French-speaking participants with DS took part in this study. The two groups were matched for nonverbal and verbal levels. We used the Benton Facial Recognition test to study the groups' ability to recognize faces, the Theory of Mind Test Revised, and the morphosyntactic production and morphosyntactic comprehension subtests from the French test battery "Instruments pour le Screening et l'Approfondissement de l'examen des DYsfonctionnements du Langage chez l'Enfant (ISADYLE)". The results of the participants with WS were systematically better than those of the participants with DS. Nevertheless, although they outperformed the participants with DS, the participants with WS were far from performing at ceiling level. Most importantly, we found no significant correlations between performances on the various standardized tests for either group of participants. The evaluation of cognitive skills in a single group of French participants with WS confirmed that they present a complex profile. Because these competences are strongly influenced by social and cultural aspects, it is therefore essential to develop studies for specific languages, including when one is examining a rare developmental disability.

Keywords: Williams syndrome; Down syndrome; cognitive profile; French; language; face recognition; theory of mind

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1. Introduction

In the last three decades, the sociocognitive profile of people with Williams syndrome (WS) has been the topic of numerous debates between defenders of nativist theories and neuroconstructivists. The performance of people with WS was initially described as atypical, as it includes relatively preserved linguistic capacities and altered nonlinguistic capacities, despite a mild to moderate intellectual disability (ID). In one of their pioneering studies of WS, Karmiloff-Smith, et al. tested the Brothers–Ring hypothesis, which posits that there is an extensive social module that is responsible for processing socially relevant stimuli, including face recognition, language, and theory of mind (ToM) [1]. They confirmed that the participants with WS in their study presented three "islet[s] of relatively preserved ability" [1] (p. 202). Nevertheless, in accordance with their previous work [2], the authors proposed a more complex interpretation of the competences observed in WS. In their view, if modules specific to a domain—in this case, language, ToM, or face recognition—actually exist, they themselves are not innate but gradually become specialized through experience on the basis of innate constraints (i.e., modularization).

Since then, many studies of WS have been carried out. The face recognition, ToM, and linguistic abilities of people with WS have been widely studied and are often compared

with the performance of people with Down syndrome (DS) (face recognition: [3,4]; ToM: [5]; language: [6–8]). In all of those studies, participants with WS consistently obtained better results than those with DS regardless of their nationality or the methodology used.

To the best of our knowledge, no study since the one by Karmiloff-Smith et al. (1995) [1] has investigated a possible link between language, ToM, and face recognition in individuals with WS. Since then, studies of neurotypical and atypical development have cast doubt on the true relationship between language and ToM [9]. A priori, ToM as a whole cannot be considered to be intact given the different results as a function of the cognitive level found in people with WS. More generally, it should be noted that several studies have found that ToM skills depend on linguistic competences [10–12]. For example, children with autism spectrum disorder (ASD) who successfully perform false belief tasks have better verbal skills than children with WS [13]. This finding led Van Herwegen et al. [14] to explain the poor performance of participants with WS on ToM tasks as being related to problems understanding narratives, rather than deficient ToM capacities, particularly because the establishment and development of language is generally delayed and clearly atypical in individuals with WS [15]. It is also very probable that these competences are strongly influenced by social and cultural aspects. In neurotypical children, for example, interlinguistic variations have been observed in early lexical [16] and morphosyntactic development [17]. Intercultural studies have also shown specific prosodic features in French-speaking vs. English-speaking children with WS [18]. In a series of studies comparing individuals with WS in Japan, the United States, Italy, and France, Reilly (2009) found evidence of a hypersocial phenotype in WS that is specific to each nationality [19].

Considered as a whole, the results of the studies conducted on people with WS, which have been obtained using very different methodologies and diverse groups of participants of different nationalities, have led to divergent conclusions concerning the capacities that are really at play in WS. These capacities are probably extremely dependent on social and cultural characteristics. Therefore, it is crucial to develop studies specific to a given language, including when one is studying a rare developmental anomaly. Consequently, this study was conducted to (1) assess the face recognition, ToM, and linguistic capacities of a single group of French speakers with WS and (2) characterize their developmental profile. We chose to compare their results with those of French-speaking participants with DS because these two genetic anomalies result in equivalent IDs and are considered as "mirror conditions", that is, a cognitive domain that is preserved in one syndrome is affected in the other and vice versa; this is true, for example, of visuospatial construction vs. linguistic skills [20]. Intersyndrome comparison has the benefit of avoiding certain difficulties related to comparisons with typically developing groups. Indeed, comparing participants with neurodevelopmental disorders who perform at ceiling level with typically developing (TD) participants who score at floor level now seems obsolete and probably contributed to overhasty conclusions regarding the preserved or altered abilities related to these syndromes (for a discussion, see [21,22], for example). In accordance with the data presented in the literature on groups of participants with WS of different nationalities, we expected that (1) individuals with WS would obtain better results in all the above-mentioned domains than those with DS; (2) if the hypothesis that a "social module" exists is valid, participants with WS should also perform well on all the tests while those with DS should perform very badly.

2. Materials and Methods

2.1. Participants

Twelve French-speaking participants with WS aged 13.0 years and 12 French-speaking participants with DS aged 15.6 years participated in this study. The participants in the WS group had positive results with the Fluorescence In Situ Hybridization (FISH) technique for 7q11.23 microdeletion and participants in the DS group had a medical diagnosis of trisomy 21. The diagnosis was confirmed by the medical teams at the institutions where these participants were being monitored. In accordance with French legislation, all participants were monitored in line with the national protocol of diagnosis and care. They were

contacted through the national Williams federation in France or through their special care homes. The participants' parents and the participants themselves consented to participate in this study. The consent statement was read in the presence of the experimenter so that she could rephrase and explain as needed.

The two groups were matched for both nonverbal and verbal performance. Nonverbal reasoning was assessed using Raven's Colored Progressive Matrices (RCPMs) [23], for which there was no difference in scores for the two groups. We opted for RCPMs rather than IQ scores. In addition to the method's ease of administration, the psychometric and developmental properties of the RCPMs mean that it is widely used with both typically developing participants and those with some kind of disorder [24,25]. Verbal skills were assessed using two tests, denomination-production and designation-lexical comprehension, from the French Instruments pour le Screening et l'Approfondissement de l'examen des DYsfonctionnements du Langage chez l'Enfant test battery (ISADYLE) [26]. The two groups did not differ significantly on these two tests. To evaluate phonological short-term memory (PSTM), we administered two other subtests from the ISADYLE battery: repetition of nonwords with consonant–vowel (CV) syllable structure and repetition of nonwords with consonant–consonant–vowel (CCV) syllable structure. The scores of the participants with WS were significantly higher than those of participants with DS on these two tests (respectively, $U = 15.5$, $p < 0.05$, and $U = 9$, $p < 0.05$).

The participants' characteristics are summarized in Table 1.

Table 1. Participants' characteristics.

	Williams Syndrome (*n* = 12, 7 Girls)		**Down Syndrome (*n* = 12, 7 Girls)**		**Max Score**
	Mean	**Range**	**Mean**	**Range**	
Chronological age	13.0	6.3–27.2	15.6	10.7–19.1	/
RCPMs	16.58	9.0–25.0	15.08	9.0–25.0	36.0
Word production	24.5	19.0–28.0	22.17	16.0–29.0	35.0
Word comprehension	26.17	21.0–28.0	24.83	21.0–28.0	28.0
CV	4.17	2.0–5.0	2.58	1.0–4.0	5.0
CCV	2.75	2.0–3.0	1.5	1.0–2.0	5.0

Note. RCPMs: Raven's Colored Progressive Matrices, CV: consonant–vowel, CCV: consonant–consonant–vowel.

2.2. Materials and Procedure

We used the computerized version of the Benton Facial Recognition Test (BFRT) [27] to study face recognition ability. This version includes 22 items divided into two parts. For the first six items, participants have to match a target photograph where the face is seen full-face to another photograph, out of a choice of six. Next, participants have to match a target photograph where the face is seen full-face with three photographs of faces shown from different angles or with different lighting.

The Theory of Mind Test Revised (ToM-Test-R) [28] is a ToM test that was initially designed for children aged 4 to 12 years with autism spectrum disorder. The test comprises 14 items divided into three blocks, which assess different levels of ToM development, namely essential prerequisites (pretending, distinction between real and imaginary, and recognition of emotions—ToM1), first-order beliefs (ToM2), and second-order beliefs (ToM3). Each block is scored out of 12, and the maximum total score for the tests is 36.

Among linguistic skills, we chose to target morphosyntactic aspects since they have been most studied in WS [15]. Morphosyntactic capacities were assessed using two subtests of the ISADYLE battery [26]: morphosyntactic production (MP) and morphosyntactic comprehension (MC). These two tests include items that require high-level general language skills and specifically evaluate morphosyntactic capacities in production and comprehension. Each test is divided into three blocks: one on simple sentences (SSP: simple sentence production; SSC: simple sentence comprehension), one on passive sentences (PSP: passive

sentence production; PSC: passive sentence comprehension), and one on tense inflections (TIP: tense inflection production; TIC: tense inflection comprehension).

All tests are standardized in French and are well designed for participants with ID. Tests were administered in a quiet room at home or in the care facility. Depending on participants' fatigue and motivation, three or four sessions were necessary to administer all the tests. The order of test presentation was randomized.

3. Results

Statistical analyses were performed with R software, version 3.2.1 [29]. Since all the scores did not respect the conditions of normality and homogeneity, we conducted nonparametric analyses (Mann–Whitney *U* test) to compare the scores obtained by the WS and DS groups. Correlation analyses were carried out using the Spearman method with a Bonferroni correction.

On average, participants with WS performed better than participants with DS on all tests (Table 2).

Table 2. Mean scores for participants with WS and DS on the three tests.

		Williams Syndrome		Down Syndrome		*p*
		Mean	**SD**	**Mean**	**SD**	
Face recognition	BFRT	37.08	6.08	30.25	2.70	***
ToM	ToMT	18.75	5.19	13.17	3.33	**
	ToM1	9.58	1.93	8.00	2.26	*
	ToM2	5.00	2.34	3.17	0.58	**
	ToM3	4.17	2.21	2.00	2.17	**
Language production	MP Total	11.25	2.63	9.75	2.67	*ns*
	SSP	8.00	2.04	6.58	1.51	**
	PSP	0.42	0.67	0.58	0.67	*ns*
	TIP	2.83	1.53	2.58	1.56	*ns*
Language comprehension	MC Total	17.33	2.10	13.67	2.71	***
	SSC	9.00	0.00	8.67	1.15	*ns*
	PSC	4.67	0.98	3.00	1.13	***
	TIC	3.25	1.36	2.00	1.41	*

Note. *ns:* not significant, *: $p < 0.05$, **: $p < 0.01$, ***: $p < 0.001$. BFRT: Benton Favial Recognition Test, ToM: theory of mind, ToMT: theory of mind total, MP: morphosyntactic production, SSP: simple sentence production, PSP: passive sentence production, TIP: tense inflection production, SSC: simple sentence comprehension, PSC: passive sentence comprehension, TIC: tense inflection production.

The WS group performed significantly better on face recognition (BFRT: $U = 18$, $p < 0.001$), total ToM score (ToMT $U = 27.5$, $p < 0.01$), and the three ToM subtests (ToM1: $U = 38$, $p < 0.05$; ToM2: $U = 29.5$, $p < 0.01$; ToM3: $U = 32$, $p < 0.01$).

For morphosyntactic competences, in comprehension (MC Tot), the overall scores for the WS group were higher than those for the DS group ($U = 20$, $p < 0.001$). Analysis of the subtests showed higher results only for PSC and TIC (PSC: $U = 20$, $p < 0.001$, and TIC: $U = 36.5$, $p < 0.05$) but not for SSC; both groups' scores on this subtest showed ceiling effects. For morphosyntactic production (MP Tot), we see the opposite pattern. Although the overall scores for the WS group are slightly higher, they are not significantly better than those of the DS group, and an analysis of the subtests shows higher scores only for SSC ($U = 31.5$, $p < 0.01$); on the other two subtests, the numbers of items produced were low, even close to 0, in the case of PSP.

The correlation analysis did not reveal any correlation between the performance on the various tests for either group (Tables 3 and 4).

Table 3. Correlations for the WS group between Benton Facial Recognition Test (BFRT), Theory of Mind Test Revised (ToMT), 'Production Syntaxique' subtest (ProdSynt), and 'Compréhension Syntaxique' subtest (CompSynt) and the 95% confidence intervals.

	ToMT	**ProdSynt**	**CompSynt**
BFRT	0.55 (−0.04 to 0.85)	−0.33 (−0.30 to 0.76)	0.39 (−0.24 to 0.79)
ToMT	-	0.18 (−0.44 to 0.68)	0.13 (−0.48 to 0.65)
ProdSynt		-	0.25 (−0.38 to 0.72)

Table 4. Correlations for the DS group between Benton Facial Recognition Test (BFRT), Theory of Mind Test Revised (ToMT), 'Production Syntaxique' subtest (ProdSynt), and 'Compréhension Syntaxique' subtest (CompSynt) and the 95 % confidence intervals.

	ToMT	**ProdSynt**	**CompSynt**
BFRT	0.24 (−0.39 to 0.71)	−0.22 (−0.70 to 0.41)	−0.02 (−0.58 to 0.56)
ToMT	-	0.35 (−0.28 to 0.77)	0.09 (−0.51 to 0.63)
ProdSynt		-	0.13 (−0.48 to 0.66)

4. Discussion

The purpose of this study was to assess face recognition, ToM, and linguistic skills in a group of French-speaking participants with WS and compare them with those of a group with DS. Given the results of previous studies, we expected that individuals with WS would obtain consistently higher scores than those with DS. This prediction was confirmed in the case of face recognition and ToM, which corroborated studies of individuals with WS of different nationalities [3,4].

As for morphosyntactic competences, the results obtained by our participants with WS are more complex to analyze.

In the case of morphosyntactic comprehension, individuals with WS generally scored higher than participants with DS. Only responses in simple sentence comprehension did not differ between the two groups, which is easy to explain by the ceiling effect found in this subtest and confirms the results of other studies [6–8,30].

In the case of morphosyntactic production, the performance of individuals with WS was superior to that of people with DS only on the simple sentence production subtest, whereas no difference was found for the subtests assessing more complex morphosyntactic structures (i.e., passive sentence and tense inflection production). This lack of significance is partially due to the fact that none of the participants in either group produced many items in a cued production situation. As such, this result is in line with studies showing that people with WS have difficulties with the most complex morphosyntactic rules (see the review by Brock, 2017) [15]. Moreover, morphosyntactic production abilities were tested with a cued production task. The problem with this kind of test is that it does not allow one to determine what level the participants' problems affect. The mediocre performance by both WS and DS groups on the passive sentences and tense inflections may be due to an actual deficit affecting the application of the rules governing these structures or to PSTM recovery issues. PSTM capacity impacts morphosyntactic abilities [31] in both neurotypical and atypical development [32,33]. In our study, we opted to use verbal protocols that may, a priori, have benefited the participants with WS and penalized those with DS, given that language problems are fairly consistently reported in the latter group [34]. That is why we were careful to match the two groups of participants for both nonverbal and verbal performance. As mentioned in the description of participants, both groups had equivalent levels of

language development. On the other hand, we found that, in the pretests, the group with WS had better PSTM than the group with DS. The better results for the individuals with WS than the group with DS in the production of simple French sentences may therefore be explained by their better PSTM, which, however, proved inadequate when the grammatical structures became more complex; French is quite complex structurally [35].

As for the hypothesis that individuals with WS have a sort of "social module", we expected our group with WS to perform successfully on all the tests and our group with DS to fail. To our surprise, the analysis of the results revealed no correlation between participants' performance on the different tests. Thus, we cannot claim that the participants with WS in our study possessed related "islets of competence", and we cannot support the Brothers–Ring hypothesis tested by Karmiloff-Smith et al. [1].

Our work did enable us to confirm that the results of French-speaking participants with WS for face recognition, ToM, and morphosyntax, as described above, were better than those of the group with DS when the two groups were matched on a verbal and nonverbal developmental level. We can also state that, in participants with IDs, high performance in one cognitive domain does not predict success in another domain.

Our study presents certain limitations. First, we chose to match participants based on both nonverbal and verbal developmental age and not chronological age. This kind of matching is not easy to apply and, although there were no statistically significant differences between the two groups on verbal and nonverbal aspects, the results still slightly favor the group with WS even though they were two years younger than the participants with DS. Second, although our sample was of a reasonable size given the rarity of WS in a country the size of France, it was still too small to allow for generalization. In addition, since the results did not respect normality due to the substantial interindividual variations, as has often been found with genetic pathologies involving ID, we were obliged to use nonparametric statistics. Future studies should examine a larger number of participants in order to verify the results.

Nevertheless, the clinical implications are still important. Although more reserved on the topic than Bellugi et al.'s (1988) [36] seminal work, research continues to present the abilities of individuals with WS in these three domains as being surprising in light of their poor performance on visuospatial tests [37], for example, and compared to other pathologies (e.g., review [15,20]). These data have probably contributed to an a priori overestimation of the theoretical performance of people with WS, by both researchers and clinicians. In France, health professionals and educators still tend to believe that the language skills of young people with WS are remarkable and overestimate their understanding and potential to learn, which can set them up for failure over time. Thus, the competences of individuals with WS are often overestimated since they are evaluated with language tests, whereas, conversely, the skills of people with DS are probably underestimated because this kind of test is used. Moreover, unlike children with WS, children with DS generally have poor PSTM performance, which affects their grammatical ability [33]. Bussy et al. (2013) [38] showed that PSTM training, using learning the self-repetition strategy, has a positive impact on lexical development and sentence comprehension in children with an ID. Since language—and communicative factors more generally—plays a fundamental role in the acquisition and development of different cognitive and social skills, it would seem to be important to assess the effects of early language training on children with DS and WS.

5. Conclusions

Despite years of research, the abilities of young people with WS are still a mystery. The capacities studied here, which are considered to be strengths in this syndrome, are probably very much influenced by sociocultural factors. Understanding and producing structurally correct and pragmatically appropriate words and sentences means that a child relies on both structural and social rules. This is equally true of the development of face recognition skills and ToM. The specific characteristics of each culture generate different results in the areas of face recognition, ToM, and language because these competences are

social in nature. It is therefore essential to develop studies for specific languages, including when one is examining a rare developmental disability.

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Institutional Review Board Statement: Ethical approval was not required for this study, according to the national and institutional guidelines. This study was carried out in accordance with the recommendations of French law that written informed consent be obtained from all subjects. All participants gave their written agreement to participate, and their parents and/or legal guardians were informed of the objectives of the study, the nature of the tasks that would be administered, and the fact that they could withdraw their agreement at any time. Their informed consent was received in writing in accordance with the Declaration of Helsinki. The medical or social and academic authorities were also informed and agreed that the students could take part, since most of the meetings took place at the educational institution.

Informed Consent Statement: Ethical approval was obtained according to the national and institutional guidelines, being the CPP (Comité de Protection des Personnes): 2022-A00341-42 for the project: ANR-20-CE28-0013. This study was carried out in accordance with the recommendations of French law. None of the participants, their parents, and/or legal guardians objected that their data would be used for research. Each was informed of the objectives of the study, the nature of the tasks that would be administered, and the fact that they could withdraw the use of their data at any time. The study was performed in accordance with the Declaration of Helsinki. Because most of the assessment meetings took place at the educational institution, the medical, social, and academic authorities were also informed and approved of the study setting.

Data Availability Statement: The data presented in this study are available on request from the corresponding author. The data are not publicly available due to ethical restrictions.

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