

# *Do infants with Down syndrome show an early receptive language advantage?*

Article

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# 1        **Do infants with Down syndrome show an early receptive language advantage?**

## 2        **Abstract**

### 3        **Purpose**

4        The study explored longitudinally the course of vocabulary and general language  
5        development in a group of infants with Down syndrome (DS) compared to a group of  
6        typically-developing (TD) infants matched on non-verbal mental ability (NVMA).

### 7        **Method**

8        We compared the vocabulary and general language trajectories of the two groups in two  
9        ways: a) at three time points during a 12 month period, and b) at 2 time points when the  
10       groups had made equal progress in non-verbal mental ability (a period of 6 months for the TD  
11       infants, versus 12 months for the infants with DS).

### 12       **Results**

13       The TD group had overtaken the DS group on all general language and vocabulary measures  
14       by the end of the 12-month period. However, expressive communication and expressive  
15       vocabulary were developing at the same rate and level in the two groups when examined over  
16       a period in which the two groups were matched in gains in non-verbal mental ability.

17       Furthermore, the infants with DS showed a receptive language advantage over the TD group;  
18       this group's auditory comprehension and receptive vocabulary scores were superior to those  
19       of the TD group at both time points when non-verbal mental ability was accounted for.

### 20       **Conclusion**

21       The results shed light on the widely reported discrepancy between expressive and receptive  
22       language in individuals with DS. Although infants with DS appear to be developing language  
23       skills more slowly than chronological age TD peers, when NVMA is taken into account,

24 infants with DS do not have expressive language delays and they seem to show a receptive  
25 language advantage.

## 26 **1. Introduction**

27 Down syndrome (DS) is the most common genetic cause of intellectual disability  
28 (Martin, Klusek, Estigarribia, & Roberts, 2009) with prevalence estimates of 1 in 691 live  
29 births (Parker et al., 2010). It results from partial or complete duplication of chromosome 21  
30 (Epstein, 1986). Characteristic features include a flat broad face, flat nasal bridge, and flat  
31 facial profile, narrow auditory canals, a small oral cavity, a relatively large tongue, and low  
32 muscle tone of the lips and tongue (Martin et al., 2009). Individuals with DS typically have  
33 an IQ of between 30 and 70 (average 50).

34 Language acquisition is delayed in DS (Roberts, Price & Malkin, 2007). Infants with  
35 DS have been reported to produce their first words at approximately 21 months (Stoel-  
36 Gammon, 2001), compared to 12 months of age for TD infants (Tomasello, 2003). First  
37 words are acquired in line with general cognitive ability (Miller, 1999). An asynchrony  
38 between receptive and expressive vocabulary has been reported for 4- to 7- year old children  
39 with DS (Caselli et al., 1998), which is similar to typically developing children (Caselli et al.,  
40 1995) in that expressive vocabulary lagged behind receptive. Expressive language in DS can  
41 be progressively delayed relative to receptive language and general non-verbal skills  
42 (Abbeduto, Warren, & Conners, 2007; Chapman & Hesketh, 2000).

43 In adolescents and adults with DS, receptive vocabulary is usually reported as a  
44 relative strength (Abbeduto, Warren & Conners, 2007) and generally in line with non-verbal  
45 mental age. Importantly, receptive vocabulary has sometimes been reported as exceeding  
46 general non-verbal abilities (Abbeduto et al., 2007; Naess et al., 2011).

47           It used to be believed that the majority of children with DS under 5 years of age have  
48 a linguistic profile characterised by receptive language skills that are in line with non-verbal  
49 mental age, and expressive language skills that are lower than expected for non-verbal mental  
50 age (Miller, 1999). Recent studies show that the picture is more complex and there are mixed  
51 findings especially when using longitudinal frameworks. Galeote et al., (2011), using a  
52 Spanish adaptation of the MacArthur–Bates CDI, reported significantly larger receptive  
53 vocabularies for 186 children with DS (aged 11 to 71 months) compared to TD children  
54 matched for mental age, while expressive vocabularies were in line with their non-verbal  
55 mental age. This study was cross-sectional and hence only provides a snapshot of  
56 development. There is a paucity of longitudinal studies and few of the existing ones have  
57 focused on language acquisition in the first three years of life with some studies focusing  
58 solely on vocabulary acquisition and others on general language acquisition. These are  
59 reviewed below.

### 60           **Longitudinal studies of vocabulary development: receptive and expressive**

61           Focusing exclusively on early acquisition of object names, Cardoso-Martins, Mervis,  
62 and Mervis (1985) followed longitudinally 6 children with DS aged 17-19 month at the start  
63 of the study, and compared them to 6 typically developing children aged 9 months at the start  
64 of the study. After an initial lack of difference between the two groups, the acquisition of  
65 object names in the DS group (comprehension and production) was reported to start to lag  
66 behind their general non-verbal cognitive skills suggesting that vocabulary acquisition  
67 develops at a slower pace than level of general cognitive abilities from an early age. Due to  
68 the very small sample size (n=6), the findings should be taken cautiously, however. A more  
69 recent study, using the Italian version of the MacArthur–Bates CDI with 18 children with DS  
70 aged between 2 and 3, Zampini and D’Odorico (2013) also reported that expressive  
71 vocabulary lags behind general cognitive development, with the main changes in vocabulary

72 development occurring at 36 months chronological age, when individual differences become  
73 more prominent. Focusing exclusively on expressive vocabulary, Te Kaat van den Os,  
74 Volman, Jongmans, and Lauteslager (2017) followed longitudinally 26 children with DS  
75 aged between 18 and 24 months at the start of the study. Parents completed the Lexi  
76 questionnaire monthly over a period of 18 months which measures expressive vocabulary and  
77 gesture use in toddlers (Schlichting & Lutje Spelberg, 2002, cited in Te Kaat van den Os et  
78 al., 2017). Wide individual variation was reported as in Zampini and D’Odorico’s study, but  
79 general cognitive abilities were related to children’s expressive vocabulary growth.  
80 Specifically, the children who made marginal progress with their vocabulary development  
81 had significantly lower general cognitive skills than the children who had a more significant  
82 growth in their vocabulary.

83 Focusing solely on receptive vocabulary, Cuskelly, Povey and Jobling (2016)  
84 investigated receptive vocabulary development from 2 years 9 months to mid adulthood in  
85 206 individuals with DS using the Peabody Picture Vocabulary Scale (PPVT). Receptive  
86 vocabulary increased up to around 20 years of age and then started to decline. The rate of  
87 receptive vocabulary development in childhood and adolescence in DS was reported to be  
88 slower than in typically developing children but there was a positive association between  
89 receptive vocabulary and general non-verbal ability.

90 In summary, the few longitudinal studies on vocabulary development suggest that, on  
91 the whole, and if we weight the findings of studies with a larger number of participants more  
92 heavily (Te Kaat van den Os et al., 2017 & Cuskelly et al., 2016), vocabulary development in  
93 children with DS is slower in the early stages of acquisition compared to typical language  
94 development and appears to be related to general cognitive abilities.

95 **Longitudinal studies of general language development: expressive language**

96 Two longitudinal studies to our knowledge have considered early expressive language  
97 development beyond vocabulary acquisition (Levy & Eilam, 2013; Oliver & Buckley, 1994).  
98 Oliver and Buckley (1994), using parental report, followed the development of vocabulary  
99 acquisition of nine children with DS (aged between 1 and 4 years) until they reached a  
100 vocabulary of 10 words, which children achieved between the ages of 19 and 38 months.  
101 Two word combinations emerged between 25 and 52 months (mean age of around 36  
102 months). Children with DS had acquired a similar number of words to TD children at the  
103 point when they started producing two word utterances. Non-verbal mental ages were not  
104 reported, hence we do not know if there was any relationship between children's language  
105 development and their non-verbal mental ability.

106 A more recent study by Levy and Eilam (2013) followed longitudinally 9 children  
107 with DS (mean age of 3 years 10 months at study entry) using a naturalistic data collection  
108 method. The children with DS were significantly delayed in entering the two-word  
109 combinations stage compared to the TD children of a similar non-verbal mental age.  
110 Specifically, while the TD children entered this stage at approximately 22 months of age, the  
111 children with DS entered this stage at approximately 55 months of age. Although the children  
112 with DS showed a typical trajectory of development over one calendar year with regard to  
113 language structure, there was atypical age of onset of two-word combinations and slower  
114 developmental pace. This deviation from typical timing was taken to suggest atypical  
115 grammatical development in children with DS. In addition, general cognitive ability was not  
116 related to the children's language status.

117 In summary, these two studies focus on expressive language only. Both agree that  
118 children with DS start producing two word combinations later than typically developing  
119 children (between 36 and 55 months of age). Moreover, Levy and Eilam (2013) propose that

120 grammatical development follows an atypical trajectory in children with DS, reflected in both  
121 a later onset and slower pace of development.

## 122 **Theoretical considerations**

123 The question of whether the developmental profiles of children with  
124 neurodevelopmental disorders can be described as ‘typical’ is a matter of considerable  
125 debate. It is unlikely that children with neurodevelopmental disorders, such as Down  
126 syndrome, would follow a typical developmental trajectory because genetic abnormalities  
127 very likely affect developmental pathways, and the adult phenotype is the product of an  
128 emergent developmental process (Karmiloff-Smith, 1998; Karmiloff-Smith, 2009; D’Souza,  
129 D’Souza & Karmiloff-Smith, 2017). Furthermore “tiny variations in the initial state” can  
130 become magnified into large domain-specific differences as a result of development  
131 (Karmiloff-Smith 1998, p. 390). Developmental timing is one parameter that influences  
132 typical development. For example, in the case of children with Down syndrome, small  
133 differences in the timing of the onset of two-word combinations (which appear in children in  
134 DS 12-24 months later than in TD children) can lead to a delay in the children’s ability to  
135 understand and produce SVO structures, which in turn can lead children to lag further behind  
136 peers in accessing relevant information in the education context. Thus, what appear to be  
137 small variations in timing in early development can compound over time, leading to a profile  
138 of severely impaired expressive language later on in adolescence and adulthood.

## 139 **Aims of current study**

140 Previous studies on language development of infants and children with DS have  
141 focused exclusively on either vocabulary, or general expressive language development.  
142 Although some studies have compared expressive and receptive vocabulary in individuals  
143 with DS, no study to our knowledge has explored the trajectories of general expressive and



144 receptive language skills (i.e. expressive and receptive communication which may or may not  
145 include grammar) and expressive and receptive vocabulary, at the early stages of language  
146 development in DS and in relation to non-verbal mental ability development. Thus, unlike  
147 most previous longitudinal studies, our study captures both the acquisition of vocabulary, and  
148 general language skills beyond single word production and comprehension in the same  
149 children, providing a more complete picture of this group of children's early language  
150 comprehension and production. It is also crucial to consider language development in infancy  
151 to understand development as it unfolds, as we cannot assume that the adult phenotype also  
152 applies to the start state of development (Karmiloff-Smith, 1998). In addition, we want to  
153 understand language within the broader context of children's general cognitive skills. The  
154 purpose of the current study is therefore twofold:

- 155 1) to establish how expressive and receptive vocabulary, and expressive and  
156 receptive general language abilities of a group of infants with DS develop over the  
157 course of 12 months in the first 3 years of life, and how their developmental  
158 trajectories compare to the language development of TD children. The two groups  
159 are compared at 3 different time points and they have equal non-verbal mental  
160 ability at Time Point 1 only (the TD group develops faster than the DS group and  
161 by Time Point 2 the TD group has higher non-verbal ability than the DS group).  
162 General language abilities are measured using a standardised assessment (the Pre-  
163 School Language Scales-4) with two components: auditory comprehension (i.e.  
164 general understanding of language) and expressive communication (general  
165 language production not restricted to grammar)
- 166 2) given that the non-verbal abilities of the TD group develop faster than the DS group,  
167 which may explain the differences in language profiles at later time points, the second aim  
168 is to establish how language development of infants with DS compares to that of TD infants

169 over a period of 12 months in which the groups have made similar progress in non-verbal  
170 mental ability. The two groups are compared at two time points and they have equal non-  
171 verbal ability at the two time points.

## 172 **2. Method**

### 173 **2.1. Ethical approval**

174 The current study was approved by the University of Reading's Research Ethics  
175 Committee and given favourable ethical opinion. TD infants were recruited from the Child  
176 Development Database at the University of Reading. This database holds the details of  
177 infants and children whose parents have consented to being contacted about studies taking  
178 place within the University of Reading. Parents of TD infants were telephoned and asked if  
179 they would be willing to take part in the study. If they were interested, then they were sent an  
180 information letter about the study and were asked to return the consent forms if they wanted  
181 to take part once they had read the information. Infants with DS were recruited through a  
182 variety of methods. Initially, the parents of infants who were taking part in language support  
183 groups at the University of Reading were sent an information letter and consent forms about  
184 the study, and were asked to get in touch, or return the consent forms if they wanted to take  
185 part. The parents of infants who were taking part in local language support groups were also  
186 approached by the experimenter and asked if they would like to take part in the study. The  
187 parents were given written and verbal information about the research study prior to testing  
188 and were informed that they were free to withdraw at any time without stating a reason.

### 189 **2.2. Participants**

190 In our original sample (see Table 1), thirty five TD infants (18 girls) were recruited  
191 into the study. All infants were being raised in a monolingual English speaking environment.

192 Thirty children with DS (12 girls) were originally recruited into the study. Three infants were  
193 exposed to languages other than English, but English was the family's dominant language.

194 Demographic data were collected for the following variables: History of Hearing  
195 Infections (Yes/No); Other Languages (Yes/No); Maternal Employment Status (Employed  
196 Full Time, Employed Part Time, Self-Employed, Unemployed, Employed but on Maternity  
197 Leave); Highest Level of Maternal and Paternal Education (None, GCSE's, A-Level, NVQ or  
198 HND, Degree, Postgraduate Degree, Other).

199 Fischer's exact tests were used to check for group differences in the demographic  
200 variables at the start of the study (Time Point 1). There were no significant group differences  
201 for Sex ( $p = .456$ ), History of ear infections ( $p = .705$ ) Maternal education ( $p = .510$ ) and  
202 Paternal education ( $p = .125$ ). A significant difference between the groups was found for  
203 Other languages used at home ( $p = .040$ ), which was due to the fact that 4 children with DS  
204 were exposed to languages other than English but English was reported to be their dominant  
205 language. In all 4 cases children were born in the UK, were attending English speaking  
206 nurseries and the parents' common language was English. A significant difference was also  
207 found for Maternal employment ( $p=.036$ ), due to fewer of the mothers of children with DS  
208 working compared to mothers of typically developing children. The data for this original  
209 sample are presented in Table 1.

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217 Table 1: Mean and standard deviation of scores at each time point for the two groups (DS and  
 218 TD) that completed each task

	TD1 (n=35) CA = 10.4 Range 9.3-11.16	DS1 (n=30) CA = 19.7 Range 17.5-23.6	TD2 (n=33) CA = 16.8 Range 16.3-17.9	DS2 (n=28) CA = 26.3 Range 23.10-30.6	TD3 (n=32) CA = 23.1 Range 22.6-24.1	DS3 (n=29) CA = 32.9 Range 30.5-36.1
NVMA	<b>32.4**</b>	<b>34.9**</b>	<b>45.1***</b>	<b>41.1***</b>	N/A	44.5
(MSEL)	<b>(sd = 2.09)</b>	<b>(sd = 4.11)</b>	<b>(sd = 3.19)</b>	<b>(sd = 5.20)</b>		(sd = 5.18)
	<b>n = 35</b>	<b>n = 30</b>	<b>n = 28</b>	<b>n = 28</b>		<b>n = 25</b>
AC (PLS-4)	<b>17.5***</b>	<b>19.7***</b>	23.4	24.2	<b>34.5***</b>	<b>28.1***</b>
	<b>(sd = 1.09)</b>	<b>(sd = 3.20)</b>	(sd = 2.25)	(sd = 2.89)	<b>(sd = 5.42)</b>	<b>(sd = 4.31)</b>
	<b>n = 35</b>	<b>n = 30</b>	n = 28	n = 28	<b>n = 31</b>	<b>n = 29</b>
EC (PLS-4)	19.1	19.1	<b>25.8***</b>	<b>23.8***</b>	<b>34.8***</b>	<b>26.3***</b>
	(sd = 2.06)	(sd = 2.85)	<b>(sd = 1.89)</b>	<b>(sd = 1.75)</b>	<b>(sd = 5.24)</b>	<b>(sd = 1.96)</b>
	n = 35	n = 30	<b>n = 28</b>	<b>n = 28</b>	<b>n = 31</b>	<b>n = 29</b>
RV (RCDI)	<b>17.9***</b>	<b>66.2***</b>	133	152	<b>344***</b>	<b>220***</b>
	<b>(sd = 20.9)</b>	<b>(sd = 51.6)</b>	(sd = 87.8)	(sd = 80.8)	<b>(sd = 112)</b>	<b>(sd = 104)</b>
	<b>n = 34</b>	<b>n = 29</b>	n = 32	n = 25	<b>n = 25</b>	<b>n = 27</b>
EV (RCDI)	<b>1.03*</b>	<b>3.38*</b>	33.3	17.8	<b>223***</b>	<b>46***</b>
	<b>(sd = 1.75)</b>	<b>(sd = 4.40)</b>	(sd = 39.4)	(sd = 22.2)	<b>(sd = 138)</b>	<b>(sd = 56.4)</b>
	<b>n = 34</b>	<b>n = 29</b>	n = 32	n = 26	<b>n = 25</b>	<b>n = 27</b>

219 CA – chronological age in months and days; TD1 – typically developing children, time point 1; TD2- typically  
 220 developing children, time point 2; TD3 – typically developing infants, time point 3; DS1- infants with Down  
 221 syndrome, time point 1; DS2- infants with Down syndrome, time point 2; DS3- infants with Down syndrome,  
 222 time point 3; NVMA – non-verbal mental ability- combined raw scores on the Visual Reception and Fine Motor  
 223 scales of the Mullen’s Scale of Early Learning (MSEL); AC – auditory comprehension; EC– expressive  
 224 communication; PLS-4 –Pre-school Language Scales-4; RV – receptive vocabulary; EV-expressive vocabulary;  
 225 RCDI-Reading Child Development Inventory; \* =  $p < .05$ ; \*\* =  $p < .01$ ; \*\*\* =  $p \leq .001$ .

### 226 2.3. Study design

227 We compared the language trajectories of the DS and TD groups in two different  
 228 ways: a) at three time points during a 12 month period, when the infants with DS were 18-20  
 229 months, 24-26 months and 30-32 months of age; and b) at 2 time points, when the two groups  
 230 had made equal progress in their non-verbal mental ability: a period of 6 months for the TD  
 231 infants, and 12 months for the infants with DS. See Table 2 below for a visual illustration of  
 232 the analysis schedule.

233 **Table 2: Study design**

COMPARISON 1 (at fixed time intervals)			
	Time Point 1	Time Point 2	Time Point 3
	CA	CA	CA
DS	Mean: 19;7	Mean: 26;1	Mean: 32;8
n=18	range:17;5-23;6	range: 24;0-30;6	range: 30;5-36;1
	CA	CA	CA
TD	Mean:10;10	Mean:16;22	Mean: 23;0
n=26	range: 9;4-11;2	range: 16;3-17;9	range 22;6-24;2
COMPARISON 2 (when groups made equal gains in NVMA scores)			
	Time Point 1	Time Point 2	
DS	CA	CA	
n=18	Mean:19;7	Mean: 32;8	
	range: 17;5-23;6	range: 30;5-36;1	
	*NVMA scores	*NVMA scores	
	mean 32.94	mean 43.55	
	range: 25-37	range 34-51	
TD	CA	CA	
n=26	Mean:10;10	Mean: 16;22	
	range: 9;4-11;2	range: 16;3-17;9	
	*NVMA scores	*NVMA scores	
	mean 32.46	mean 45.34	
	range 30-37	range 38-55	

234 Note: CA-chronological age in months and days; NVMA-non-verbal mental age;  
 235 \*NVMA scores are derived by summing the Visual Recognition and Fine Motor Skills scores

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237 At Time Point 1, the DS group had significantly higher non-verbal mental ability as measured  
238 by the Mullen Scale of Early Learning (Mullen, 1995) than the TD group, [ $t(1, 41.540) = -$   
239  $2.975, p=0.05$ ]. To match the groups on non-verbal mental ability at the first point of  
240 measurement, we first excluded cases with missing data for either non-verbal mental ability  
241 or general language measures (Pre-School Language Scale data, see below) at Time Points 1,  
242 2 or 3. This left 26 typically-developing infants, and 23 infants with DS who had completed  
243 non-verbal mental ability and language measures at all three time points. An independent  
244 samples t-test revealed that these groups were not significantly different in terms of NVMA  
245 at Time Point 1:  $t(30.8) = -1.98, p = .057$ . However, Mervis & Robinson (2003) recommend  
246 that groups cannot be assumed to be matched on a control variable unless a  $p$  value of at least  
247 0.50 is found in the test of group differences. In addition to this, according to Piaggio,  
248 Elbourne, Altman, Pocock and Evans, (2006), groups are matched if there is an adequately  
249 small effect size “which might be defined as the smallest value at which a difference in  
250 groups would be clinically meaningful” (Piaggio et al., 2006 in Kover & Atwood, p.6). Rubin  
251 (2001) proposed that the standardized mean difference be close to zero (less than half a  
252 standard deviation apart;  $d \leq .5$ ). We therefore further removed the highest scoring  
253 participants with DS until the groups were matched by this criterion. Thus the final sample  
254 has 26 typically-developing infants and 18 infants with DS matched for non-verbal mental  
255 ability:  $t(24.7) = -.567, p = .576$ , Cohen’s  $d = 0.15$  (see Table 3 for matched group  
256 comparisons on all measures).

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262 Table 3: Mean and standard deviation of scores at each time point for the two matched groups  
 263 (DS and TD) that completed each task when matched for NVMA at Time Point 1

	TD1 (n=26) CA = 10.1	DS1 (n=18) CA = 19.2	TD2 (n=26) CA = 16.2	DS2 (n=18) CA = 26.1	TD3 (n=26) CA = 23	DS3 (n=18) CA = 32.8
NVMA (MSEL)	32.5 (sd = 1.86)	32.9 (sd = 3.26)	<b>45.3***</b> <b>(sd = 3.20)</b>	<b>38.9***</b> <b>(sd = 3.08)</b>	N/A	43.6 (sd = 4.98)
AC (PLS-4)	<b>17.4***</b> <b>(sd = 1.14)</b>	<b>19.2***</b> <b>(sd = 2.05)</b>	23.5 (sd = 2.32)	23.6 (sd = 2.33)	<b>34.2***</b> <b>(sd = 5.48)</b>	<b>27.0***</b> <b>(sd = 3.40)</b>
EC (PLS-4)	18.8 (sd = 1.89)	18.3 (sd = 2.22)	<b>25.6**</b> <b>(sd = 1.86)</b>	<b>23.8**</b> <b>(sd = 1.73)</b>	<b>34.9***</b> <b>(sd = 5.21)</b>	<b>25.6***</b> <b>(sd = 1.58)</b>
RV (RCDI)	<b>18.3**</b> <b>(sd = 20.7)</b>	<b>41.5**</b> <b>(sd = 31.8)</b>	137 (sd = 91.2)	124 (sd = 66.8)	<b>347***</b> <b>(sd = 107)</b>	<b>190***</b> <b>(sd = 94.8)</b>
EV (RCDI)	0.77 (sd = 1.14)	2.83 (sd = 4.68)	32.9 (sd = 42.1)	18.1 (sd = 24.1)	<b>228***</b> <b>(sd = 139)</b>	<b>32.1***</b> <b>(sd = 36.3)</b>

264 CA – chronological age in months and days; TD1 – typically developing children, time point 1; TD2- typically  
 265 developing children, time point 2; TD3 – typically developing infants, time point 3; DS1- infants with Down  
 266 syndrome, time point 1; DS2- infants with Down syndrome, time point 2; DS3-infants with Down syndrome,  
 267 time point 3; NVMA – non-verbal mental ability- combined raw scores on the Visual Reception and Fine Motor  
 268 scales of the Mullen’s Scale of Early Learning (MSEL); AC – auditory comprehension; EC– expressive  
 269 communication; PLS-4 –Pre-school Language Scales-4; RV – receptive vocabulary; EV-expressive vocabulary;  
 270 RCDI-Reading Child Development Inventory; \* =  $p < .05$ ; \*\* =  $p < .01$ ; \*\*\* =  $p \leq .001$ .

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272 For these matched groups, the TD infants (13 girls, 13 boys) had a mean age of 10  
 273 months and 10 days (range 9 months 4 days-11 months 2 days) at Time Point 1; 16 months  
 274 and 22 days (range 16 months 3 days-17 months 9 days) at Time Point 2; and 23 months  
 275 (range 22 months 6 days-24 months 2 days) at Time Point 3.

276 The children with DS (7 girls, 11 boys) had a mean age of 19 months and 18 days  
277 (range 17 months 5 days-23 months 6 days) at Time Point 1; 26 months and 10 days (range  
278 24 months- 30 months 6 days) at Time Point 2; and 32 months and 8 days (range 30 months 5  
279 days-36 months 1 day) at Time Point 3. Of these, 2 infants were exposed to languages other  
280 than English, but English was the family's dominant language.

281 Fischer's exact tests were used to check for group differences in the demographic  
282 variables. There were no significant group differences for Sex ( $p = .547$ ), History of ear  
283 infections ( $p = .409$ ), Maternal education ( $p = .666$ ), Paternal education ( $p = .511$ ) and  
284 Paternal employment ( $p=.162$ ). A significant difference was found for Maternal employment  
285 ( $p=.024$ ), due to the fact that fewer of the mothers of children with DS were working  
286 compared to mothers of typically developing children. However, because there were no  
287 differences in maternal education, maternal employment status was not used as an exclusion  
288 criterion.

## 289 **2.4. General Procedure**

### 290 **2.4.1. Language and non-verbal measures**

291 At the three time points, infants were administered the same set of measures of their  
292 receptive and expressive general language, expressive and receptive vocabulary and non-  
293 verbal mental ability. The measures are described below.

#### 294 ***Mullen Scales of Early Learning (MSEL)***

295 Non-verbal mental ability was assessed using the MSEL (Mullen, 1995), a  
296 standardised measure of cognitive functioning for infants aged 0-68 months. Two of the five  
297 scales were administered: the Visual Reception scale and the Fine Motor scale. The Visual  
298 Reception Scale tests the infant's visual discrimination and visual memory, and requires the  
299 skills of visual organisation, visual sequencing, and visual spatial awareness, including



300 concepts of size, shape, and position. The Fine Motor Scale provides a measure of visual-  
301 motor ability. The items on this subscale require visually-directed motoric planning, and  
302 primarily assess unilateral and bilateral manipulation. Non-Verbal Mental Ability scores were  
303 derived by combining the raw scores of the Visual Reception and Fine Motor scales. T-scores  
304 were not used, as most infants with DS obtained the lowest possible value (20), masking the  
305 variability in their raw scores. Converting to T-scores would also make group comparisons  
306 meaningless, due to the differences in the groups' chronological ages.

307 ***Preschool Language Scales-4 (PLS-4)***

308 The PLS is a standardised assessment composed of two subscales: Auditory  
309 Comprehension and Expressive Communication. The Auditory Comprehension subscale  
310 evaluates understanding of language and the Expressive Communication subscale was used  
311 to determine how well children communicated with others, vocally and socially. Please note  
312 that these two measures of general receptive and expressive language do not exclusively  
313 focus on grammar. Receptive and expressive language scores were derived from the raw  
314 scores on the Auditory Comprehension and the Expressive Communication subscales  
315 respectively. Standardised scores were not used, as standardised scores for the infants with  
316 DS were often the lowest possible value (55), masking the variability in infants' raw scores.  
317 Converting raw scores to standardised scores would have made group comparisons of  
318 language abilities meaningless, due to the differences in the groups' chronological ages. Use  
319 of raw scores is common in the literature on atypical populations (for example: Klein &  
320 Mervis, 1999; Mason-Apps et al., 2018; Seager et al., 2018, van Herwegen, Tim, Smith &  
321 Dimitriou, 2015).

322 ***Reading Communicative Development Inventory (Reading CDI)***

323 Receptive and Expressive Vocabulary scores were measured using the Reading  
324 Communicative Development Inventory (CDI), an adaptation of the Oxford CDI (Hamilton,

325 Plunkett, & Schafer, 2000). This is a parental report measure, comprising a checklist of words  
326 that a child might know, in 20 semantic categories, and additional sections to indicate use of  
327 word endings, word forms, and sentences. Parents were sent the checklist to complete at  
328 home in the week prior to their visit. Parents were asked to indicate which words their child  
329 understood (but did not say), which words the child said, and which words their child both  
330 understood and said. Parents of infants with DS were also asked to indicate which words the  
331 child both understood and produced signs for. Receptive Vocabulary scores were derived  
332 from the number of words parents indicated that the child understood or understood and said.  
333 Expressive Vocabulary scores were derived from the number of words that the parents  
334 indicated that the child understood and said. Signs were excluded from the calculation of  
335 scores.

### 336 **3.0. Results**

337 To address the first aim of the study, first we present between group comparisons at each  
338 time point (TP1, TP2 and TP3), for the TD children and participants with DS matched on  
339 NVMA at TP1. Then, in order to address the second aim of the study, we compare the  
340 language development of the two groups over a period when they have made similar gains in  
341 non-verbal mental ability (which is at two time points only, i.e. Time Point 2 for the TD  
342 group and Time Point 3 for the DS group)

#### 343 **3.1. Between group comparisons at each time point of testing (to address aim 1)**

344 To address the first aim of the study, we first present comparisons between the TD  
345 children and participants with DS (matched on NVMA at TP1) at each time point (TP1, TP2  
346 and TP3). A 2 x 3 mixed design ANOVA was run in each analysis, with Group as between-  
347 subjects variable, and Time as a within-subjects variable. Table 3 above shows the raw scores  
348 for each group on all the measures collected at each time point. Significant group differences  
349 are marked with an asterisk.

350

INSERT TABLE 3 AROUND HERE

351

### 3.1.1 Auditory comprehension

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Figure 1 shows the mean Auditory Comprehension (AC) scores for the two groups at TP1, TP2 and TP3. The ANOVA found a main effect of Time, with AC scores increasing at each Time Point,  $F(1.30,51.8) = 231.366, p < .001 (\eta^2=.853)$ . There was a main effect of Group,  $F(1,40) = 5.631, p = .023 (\eta^2=.123)$ , and a significant Group x Time interaction,  $F(1.30,51.8) = 35.268, p < .001 (\eta^2=.469)$ .

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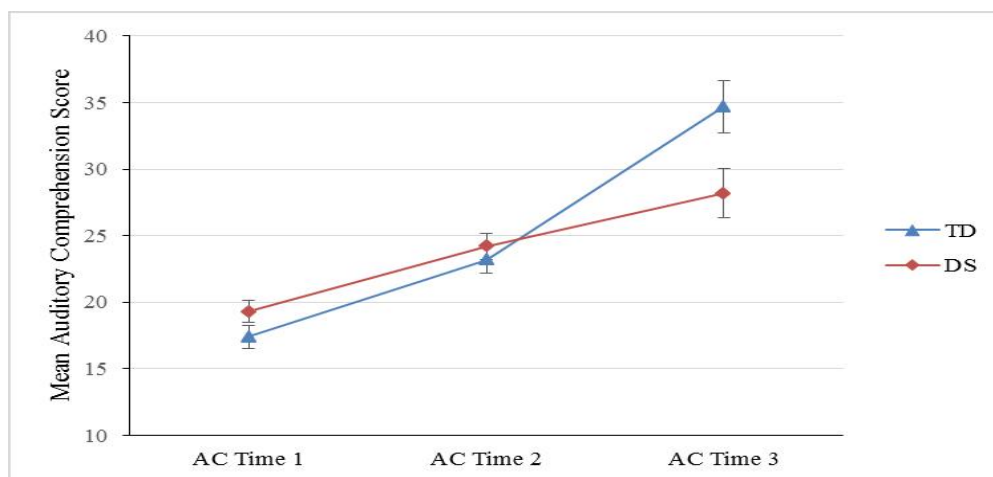
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362

Simple main effects analysis revealed that both the TD and DS group made significant gains in AC scores at each time point (all  $ps < .001$ ). The DS group had significantly higher AC scores than the TD group at TP1,  $F(1,40) = 12.566, p = .001 (\eta^2=.239)$ , there were no significant differences between the DS and TD group at Time Point 2,  $F(1,40) = 0.205, p = .654 (\eta^2=.005)$ , and the TD had significantly higher AC scores compared to the DS group at Time Point 3,  $F(1,40) = 24.130, p < .001 (\eta^2=.376)$ .

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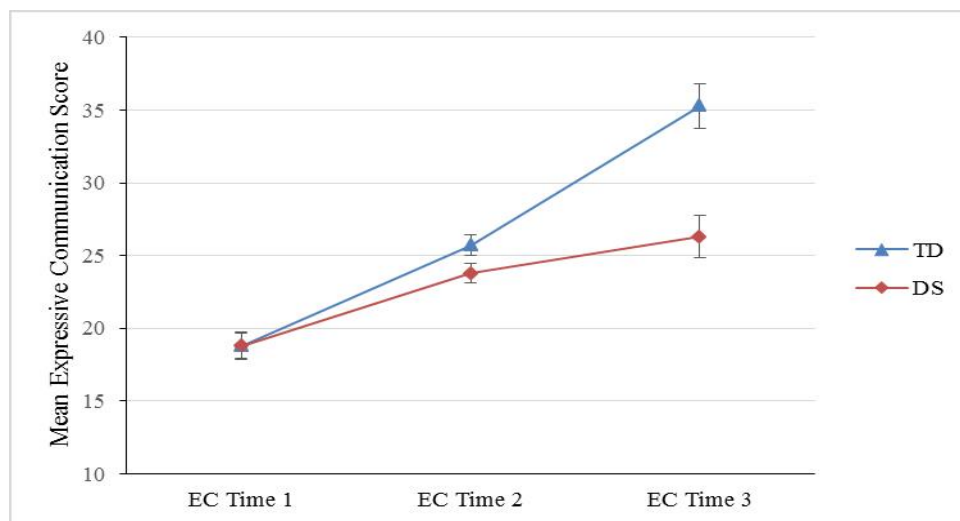
**Figure 1.** Mean Auditory Comprehension scores for the TD and DS groups at Time Point 1, Time Point 2, and Time Point 3

366

### 3.1.2. Expressive Communication

367 Figure 2 shows the mean Expressive Communication (EC) scores for the TD and DS  
368 groups at TP1, TP2 and TP3. The ANOVA found a main effect of Time,  $F(1.41,56.4) =$   
369  $310.477, p < .001 (\eta^2=.886)$ , with EC scores increasing at each Time Point. There was a  
370 significant main effect of Group,  $F(1,40) = 30.814, p < .001 (\eta^2=.435)$ , and a significant  
371 Group x Time interaction,  $F(1.41,56.4) = 50.843, p < .001 (\eta^2=.560)$ .

372 Simple main effects analysis revealed that both the TD and DS group made  
373 significant gains in EC scores at each time point (for the TD group, all  $ps < .001$ ; for the DS  
374 group TP1 andTP2,  $p < .001$ , and for TP2 andTP3,  $p = .019$ ). The analysis also showed that  
375 there were no significant differences between the TD and DS group at Time Point 1,  $F(1,40)$   
376  $= 0.991, p = .325 (\eta^2=.024)$ . However, the TD group had significantly higher EC scores than  
377 the DS groups at TP 2,  $F(1,40) = 9.308, p = .004 (\eta^2=.189)$ , and Time Point 3,  $F(1,40) =$   
378  $53.485, p < .001 (\eta^2=.572)$ .



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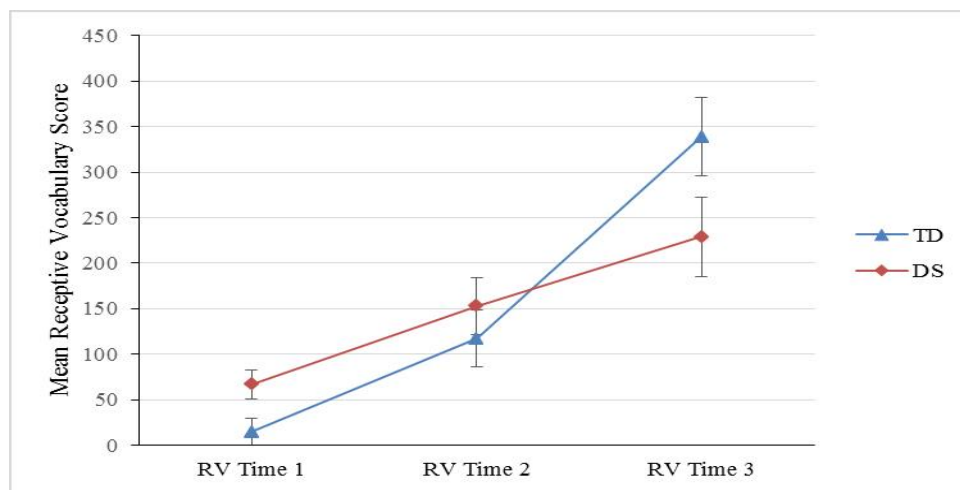
380 **Figure 2.** Mean Expressive Communication scores for the TD and DS groups at Time Point 1, Time  
381 Point 2, and Time Point 3

### 382 3.1.3. Receptive Vocabulary

383 Figure 3 shows the mean Receptive Vocabulary (RV) scores for the typically-  
384 developing group (TD) and the group of infants with Down syndrome (DS) at TP1, TP2 and  
385 TP3. Data is only presented for those participants for whom RV data was available at all

386 three time points (for the typically-developing group, N=21; for the group of infants with DS,  
387 N=17). The ANOVA found a main effect of Time, with RV scores increasing at each Time  
388 Point,  $F(1.45,52.2) = 209.392, p < .001 (\eta^2=.853)$ . There was a significant main effect of  
389 Group,  $F(1,36) = 4.593, p = .039 (\eta^2=.113)$ , and a significant Group x Time interaction,  
390  $F(1.45,52.2) = 33.350, p < .001 (\eta^2=.481)$ .

391 Simple main effects analysis revealed that both the TD and DS group made  
392 significant gains in RV scores at each time point (all  $ps < .001$ ). The analysis also showed  
393 significantly higher RV scores for the DS than the TD group at Time Point 1,  $F(1,36) =$   
394  $10.497, p = .003 (\eta^2=.226)$ , no significant differences between the DS and TD group at Time  
395 Point 2,  $F(1,36) = 0.005, p = .945 (\eta^2<.001)$ , significantly higher RV scores for the TD group  
396 compared to the DS group at Time Point 3,  $F(1,36) = 21.024, p < .001 (\eta^2=.369)$ .



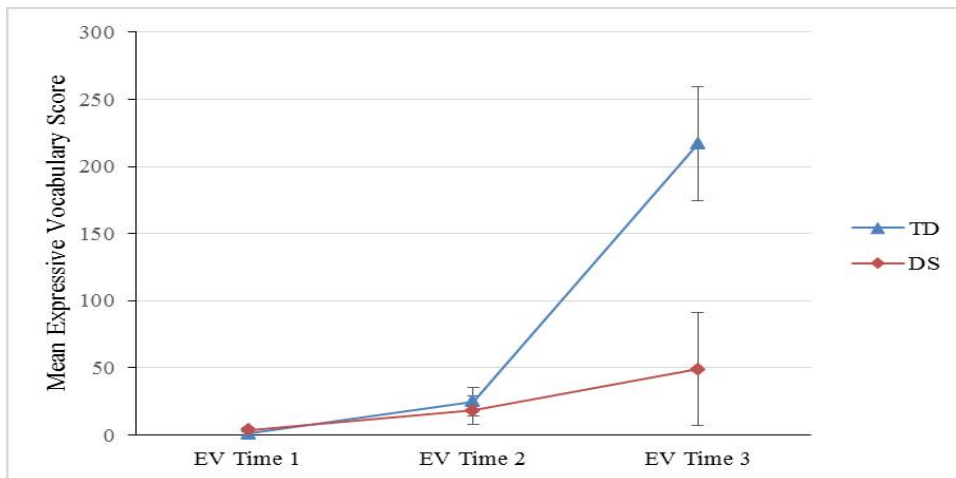
397  
398 **Figure 3.** Mean Receptive Vocabulary scores for the TD and DS groups at Time Point 1, Time Point 2, and  
399 Time Point 3

#### 400 3.1.4. Expressive Vocabulary

401 Figure 4 shows the mean Expressive Vocabulary (EV) scores for the typically-  
402 developing group (TD) and the group of infants with Down syndrome (DS) at TP1, TP2 and  
403 TP3. Data is only presented for those participants for whom Expressive Vocabulary data was  
404 available at all three time points (for the TD group, N = 21, for the DS group, N = 17). The

405 ANOVA found a main effect of Time, with EV scores increasing at each Time Point,  
406  $F(1.08,38.9) = 53.681, p < .001 (\eta^2=.599)$ . There was a significant main effect of Group,  
407  $F(1,40) = 23.239, p < .001 (\eta^2=.392)$ , and a significant Group x Time interaction,  
408  $F(1.08,38.9) = 34.123, p < .001 (\eta^2=.487)$ .

409 Simple main effects analysis revealed that the TD group made significant gains in EV  
410 scores at each time point (all  $ps < .001$ ). For the DS group, the gain in EV scores between  
411 TP1 and TP2 was significant ( $p = .030$ ), but the gain between TP2 and TP3 was not ( $p =$   
412  $.481$ ). There were no significant differences between the TD and DS groups at TP1,  $F(1,36) =$   
413  $3.609, p = .066 (\eta^2=.091)$ , or TP2,  $F(1,36) = .612, p = .439 (\eta^2=.017)$ , but the TD group had  
414 significantly higher EV scores than the DS group at TP3,  $F(1,36) = 31.372, p < .001$   
415 ( $\eta^2=.466$ ).



416  
417 **Figure 4.** Mean Expressive Vocabulary scores for the TD and DS groups at Time Point 1, Time Point 2, and  
418 Time Point 3

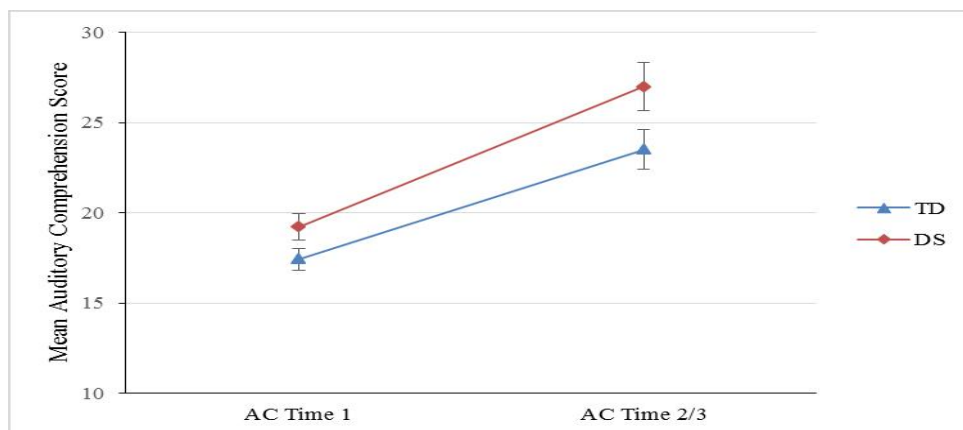
419  
420 **3.2. Language development when the two groups are matched on growth in Non-**  
421 **Verbal Mental Ability (to address aim 2 of the study)**

422 To address the second aim of the study, we compared the language development of the two  
423 groups over a period when they had made similar gains in non-verbal mental ability (i.e. from  
424 TP 1 to TP 2 for the TD group and from TP 1 to TP 3 for the DS group). This was a period of

425 12 months for the DS group and 6 months for the TD group. At the final time points included,  
426 the two groups did not differ in NVMA,  $t(1,42)=1.45$ ,  $p=.154$ , Cohen's  $d = 0.42$ . For each  
427 analysis, we ran a mixed-design ANOVA, with Group (TD, DS1) as a between-subjects  
428 variable, and Time (TP1 and either TP2 or TP3 depending on group) as a within-subjects  
429 variable.

### 430 3.2.1 Auditory Comprehension

431 Figure 5 shows the mean AC scores for the TD group (at Time Point 1 and 2) and the  
432 DS group (at Time Point 1 and 3). There was main effect of Time, with AC scores increasing  
433 between the two time points,  $F(1,42) = 245.734$ ,  $p < .001$  ( $\eta^2 = .854$ ). The ANOVA found a  
434 significant main effect of Group,  $F(1,42) = 24.173$ ,  $p < .001$  ( $\eta^2 = .365$ ), but the Group x  
435 Time interaction was not significant  $F(1,42) = 3.703$ ,  $p = .061$  ( $\eta^2 = .081$ ). Infants with DS  
436 had significantly better auditory comprehension than matched TD peers.



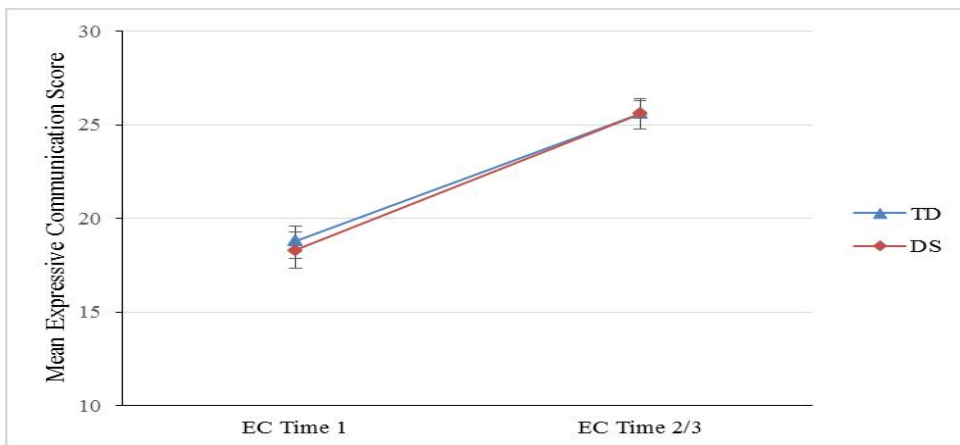
437

438 **Figure 5.** Mean Auditory Comprehension scores for the typically-developing group at Time Point 1 and Time  
439 Point 2, and for the group of infants with Down syndrome at Time Point 1 and Time Point 3, when groups were  
440 matched for NVMA

### 441 3.2.2. Expressive Communication

442 Figure 6 shows the mean Expressive Comprehension (EC) scores for the typically-  
443 developing group (at Time Point 1 and 2) and the group of infants with Down syndrome (at  
444 Time Point 1 and 3). The ANOVA found a main effect of Time, with Expressive

445 Communication scores increasing between the two time points (TP1 and TP2 for the TD  
446 group and TP1 and TP3 for the DS group),  $F(1,42) = 567.289, p < .001 (\eta^2 = .931)$  for both  
447 groups. There was no main effect of Group,  $F(1,42) = 0.328, p = .570 (\eta^2 = .008)$ , and no  
448 significant Group x Time interaction,  $F(1,42) = 0.908, p = .346 (\eta^2 = .021)$ . The Expressive  
449 Communication skills of children with DS were in line with those of their NVMA growth-  
450 matched peers.



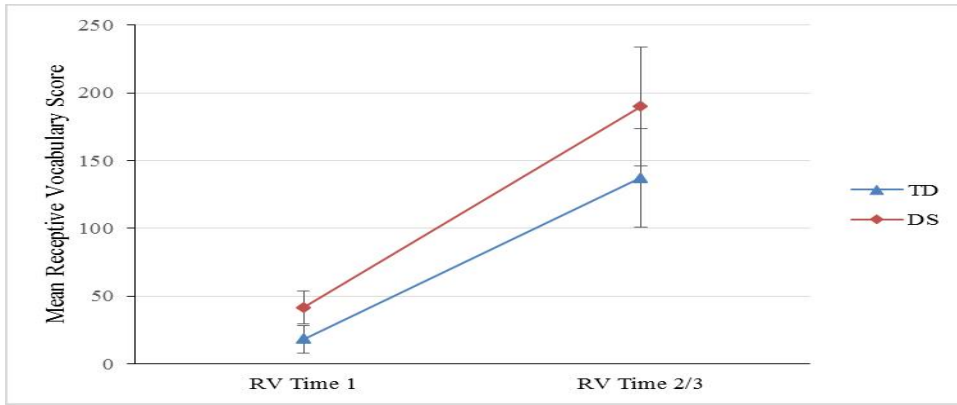
451

452 **Figure 6.** Mean Expressive Communication scores for the typically-developing group at Time Point 1 and Time  
453 Point 2, and for the group of infants with Down syndrome at Time Point 1 and Time Point 3, when groups were  
454 matched for NVMA

### 455 3.2.3. Receptive Vocabulary

456 Figure 7 shows the mean RV scores for the TD group (at Time Point 1 and 2) and for  
457 the DS group (at Time Point 1 and 3). There was a main effect of Time, with RV scores  
458 increasing between the two time points,  $F(1,42) = 11.940, p < .001 (\eta^2 = .739)$ . The ANOVA  
459 found a main effect of Group,  $F(1,42) = 5.033, p = .030 (\eta^2 = .107)$ , but no significant Group  
460 x Time interaction,  $F(1,42) = 1.441, p = .237 (\eta^2 = .033)$ . Infants with DS had significantly  
461 larger receptive vocabularies than their matched TD counterparts.





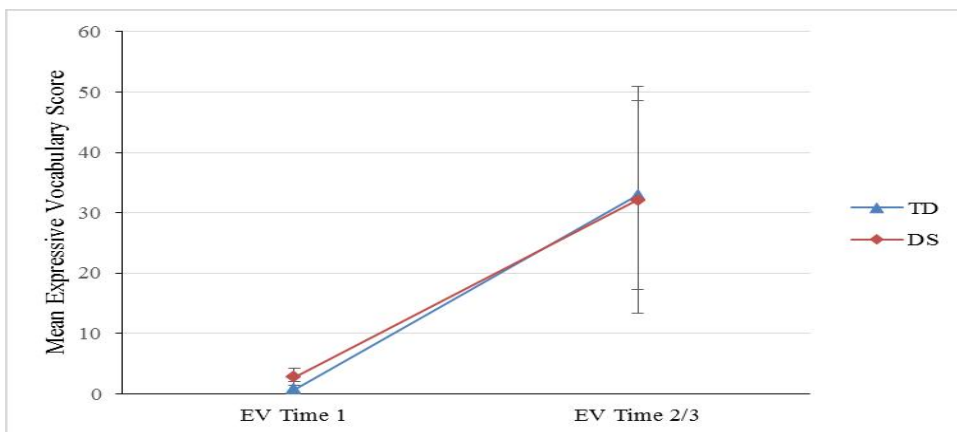
462

463 **Figure 7.** Mean Receptive Vocabulary scores for the typically-developing group at Time Point 1 and Time Point  
 464 2, and for the group of infants with Down syndrome at Time Point 1 and Time Point 3, when groups were  
 465 matched for NVMA

466

467 *3.2.4. Expressive Vocabulary*

468 Figure 8 shows the mean EV scores for the TD group (at TP1 and TP2) and for the  
 469 DS group (at TP1 and TP3). There was a main effect of Time, with EV scores increasing  
 470 significantly between the two time points,  $F(1,42) = 26.000, p < .001$  ( $\eta^2 = .382$ ). The  
 471 ANOVA found no main effect of Group,  $F(1,42) = 0.009, p = .924$  ( $\eta^2 < .001$ ), and no  
 472 significant Group x Time interaction,  $F(1,42) = 0.059, p = .809$  ( $\eta^2 = .001$ ).



473

474 **Figure 8.** Mean Expressive Vocabulary scores for the typically-developing group at Time Point 1 and Time  
 475 Point 2, and for the group of infants with Down syndrome at Time Point 1 and Time Point 3, when groups were  
 476 matched for NVMA

477

478

#### 479 **4.0. Discussion**

480 The purpose of this paper was to investigate the course of language development in a  
481 group of infants with DS compared to a group of TD infants. Specifically, we wanted to find  
482 out how language develops over the course of one calendar year after an initial time point at  
483 which groups did not differ in terms of non-verbal abilities. The second aim was to compare  
484 the trajectories of language development shown by the two groups across a period in which  
485 the groups made equal progress in their non-verbal mental ability (6 months for the TD  
486 infants; 12 months for the infants with DS).

487 The results of the whole groups' analyses, which was the first aim of the study,  
488 showed that, although the DS group were ahead of the TD group on auditory comprehension  
489 and receptive vocabulary at Time Point 1, by Time Point 2 (a period of 6 months), the TD  
490 group has already caught up with the DS group on auditory comprehension and receptive  
491 vocabulary, and significantly outperformed the DS group on expressive communication. By  
492 Time Point 3, which was a period of 12 months, the TD group significantly outperformed the  
493 DS group on all language and vocabulary measures. These findings are in line with other research  
494 studies which have shown that language in children with DS develops more slowly than in  
495 typically developing children (Abbeduto, Warren & Conners, 2007; Levy & Eilam, 2013).  
496 Inspection of the trajectories in our study seems to suggest that children with DS have more  
497 delays in expressive than in receptive language. For example, while TD children are reported  
498 to produce an average of 228 words by Time Point 3, children with DS are reported to  
499 produce on average of 32 words. With regard to receptive vocabulary, on the other hand,  
500 children with DS are reported to understand on average 190 words compared to 347 for the  
501 TD children, which is a smaller discrepancy between the TD and DS groups compared to  
502 expressive vocabulary. Such results suggest potential strengths with regard to receptive

503 vocabulary and are in line with what has already been reported about a possible receptive  
504 vocabulary advantage (Abbeduto et al., 2007; Caselli et al., 1998; Miller et al., 1999).

505         The second aim of the study was to compare the trajectories of language development  
506 between the two groups across a period in which the groups had made equal progress in their  
507 non-verbal mental ability. Our findings show that, once the language abilities were compared  
508 when both the TD and DS groups had made equal gains in terms of non-verbal mental ability  
509 development, both expressive communication and expressive vocabulary showed the same  
510 rate and level of development for the infants with DS as for the TD infants. Importantly,  
511 receptive vocabulary and auditory comprehension were significantly higher for the DS group  
512 compared to the TD group. Furthermore, for all four language measures, the trajectory of  
513 development in the DS group was very close to that of the TD group. There were no  
514 interactions between Time and Group in any analysis, showing that early expressive language  
515 in infants with DS seems to be developing entirely in line with their non-verbal mental  
516 abilities. This pattern was consistent for the two expressive (both vocabulary and general  
517 expressive communication) and two receptive assessments (vocabulary and general auditory  
518 comprehension). The data show that when non-verbal mental ability is taken into account,  
519 expressive communication and expressive vocabulary in the children with DS seem to be  
520 comparable to the TD group. On the other hand, both auditory comprehension and receptive  
521 vocabulary scores in the DS group were above those of the TD group, suggesting that our  
522 group of infants with DS may display a receptive language advantage at both time points  
523 relative to their non-verbal ability. This finding is in line with the findings of Galeote,  
524 Sebastian, Cheka, Rey and Soto (2011) for Spanish speaking children with Down syndrome  
525 who also reported that expressive vocabulary did not lag behind non-verbal mental ability,  
526 and that the receptive vocabulary of infants with DS was larger than that of mental age  
527 matched controls. Our findings, however, do not fully support those of Zampini and

528 D’Odorico (2013) who used the Italian version of the MacArthur–Bates Communicative  
529 Development Inventories and found significant differences in productive vocabulary size  
530 between children with DS and typically developing developmental age matched controls.  
531 However, it should be pointed out that the children with DS in the Zampini and D’Odorico’s  
532 study were older at the first point of measurement (they had a developmental age of 18  
533 months at the first time point whereas the infants with DS in our study had a non-verbal  
534 mental age of 9-10 months at the first time point). Thus our study captures the earliest stages  
535 of language acquisition in DS and shows that, at the point when expressive vocabulary and  
536 expressive communication emerge, children with DS are likely to be no different from  
537 typically developing children of a similar non-verbal mental ability.

538 **4.1. Do expressive and receptive language in infants with Down syndrome**  
539 **develop atypically compared to neurotypical infants?**

540 The data from our study suggest that when infants with Down syndrome are in the  
541 pre-linguistic and early stages of linguistic development, i.e. between 18 and 32 months of  
542 age, they seem to be delayed only to the extent expected given their non-verbal mental  
543 ability. This suggests that the language of infants with DS may not yet be developing  
544 atypically compared to neuro-typical infants. Importantly, our group of infants with Down  
545 syndrome: 1) did not seem to show any expressive language deficits, relative to their non-  
546 verbal mental ability, when compared to the typically-developing group at Time Point 1; and  
547 2) showed a relative strength in receptive vocabulary and auditory comprehension compared  
548 to TD infants of a similar non-verbal mental ability.

549 It is generally accepted that a discrepancy between receptive and expressive language  
550 skills is characteristic of the typical adult phenotype for individuals with Down syndrome.  
551 Hence, one could argue that the widely-reported relative strengths in receptive language  
552 abilities (including both general understanding of language and receptive vocabulary) are

553 present in Down syndrome from early on in development, mirroring the adult phenotype.  
554 This would be too simplistic an explanation.

555         The picture is more complex because the infants with Down syndrome in our study  
556 did not show any deficits in expressive language skills relative to non-verbal mental ability  
557 when compared to the typically-developing infants at Time Point 1. At Time Point 3, their  
558 expressive language skills (including expressive vocabulary and expressive communication)  
559 were in line with their non-verbal mental abilities. On the basis of the adult Down syndrome  
560 phenotype, one would expect expressive language (expressive vocabulary and/or general  
561 expressive communication) to be lagging behind non-verbal mental ability (Abbeduto et al.,  
562 2007; Chapman, Seung, Schwartz, & Kay-Raining Bird, 1998). This was not the case with  
563 our findings. The reason for this may be that our control group were very young at the first  
564 time point (between 9-10 months of age), when infants are still predominantly babbling and  
565 do not produce language as such. Thus, when compared to infants with Down syndrome aged  
566 18-22 months who were also predominantly still in the babbling stage, no differences in  
567 expressive language were evident between children with DS and neurotypical children.  
568 Importantly, however, the expressive language skills in the Down syndrome group appeared  
569 to be following the same developmental trajectory as the typically-developing group. It is  
570 likely that we did not find any differences between the groups because neither group had  
571 started using grammar yet (in terms of combining two words/morphemes). In the Levy and  
572 Eilam (2013) study, it was the onset of combinatorial language (i.e. the onset of combining  
573 two morphemes together) which was very delayed for some young children with DS.

574         The fact that an early expressive language deficit was not apparent at this early stage  
575 of development suggests that the later (and finally adult) DS language phenotype may emerge  
576 as a function of development. Deficits in expressive language skills relative to receptive  
577 language and general non-verbal mental ability in individuals with DS become more obvious

578 once two-word combinations increase (Chapman, Hesketh & Kistler, 2002). A limitation of  
579 the current study is the fact that the groups were only followed for one year after the initial  
580 time point. At the final time point, the TD group had a mean age of 23 months and the group  
581 of infants with DS had a mean age of 32 months. Because the infants were quite young at the  
582 final time point, both groups were still in the very early stages of language acquisition, with  
583 some participants having not even advanced to combining two words. Since it is expressive  
584 language and syntax development that are highlighted as particular areas of difficulty for  
585 individuals with DS, especially in later childhood and adulthood, it would be informative if  
586 the infants from this study were followed up at a later stage, when relative difficulties with  
587 grammar may have become more apparent.

588         Despite the limitations of studying infants in the earliest stages of language  
589 development, by comparing infants with DS to neuro-typical infants of a similar non-verbal  
590 mental ability, we were able to reveal potential strengths and weaknesses in the early  
591 language phenotype for individuals with DS. This has both theoretical and clinical  
592 implications.

#### 593         **4.2. Theoretical and clinical implications**

594         From a theoretical perspective, by taking a development approach and by accounting  
595 for development in other aspects of cognition (not exclusively focusing on language), we  
596 were able to characterise the earliest stages of language development in infants with Down  
597 syndrome and show that there may be an early receptive language advantage. In addition, the  
598 onset of expressive language (in terms of productive expressive vocabulary and expressive  
599 communication) at this initial stage of acquisition seems to be as expected for the level of  
600 non-verbal mental ability. However, our study also shows that non-verbal abilities in infants  
601 with DS may have a delayed onset and pace of development compared to neuro-typical

602 infants. This has also been shown in other studies of cognitive development of young  
603 children with DS studied longitudinally at 12 and 30 months showing that infants with Down  
604 syndrome make fewer gains in overall cognitive skills than children with other  
605 neurodevelopmental disorders matched on mental age (Fidler, Most, Booth-LaForce & Kelly,  
606 2008). Although these delays may appear small at this early point in development, we know  
607 that small differences in developmental timing (in this case of the acquisition of general  
608 cognitive skills) can impact on language development over time and result in more obvious  
609 deficits in phenotypic outcomes (Annaz, Karmiloff-Smith & Thomas, 2008). When the onset  
610 of development of a particular skill is delayed and not in line with the typical developmental  
611 timing, there may be cascading effects later on (Marsten & Cicchetti, 2010). Existing  
612 research suggests that non-verbal mental abilities/general cognitive skills are related to, and  
613 can account for, language development (Casby, 1992). For example, research in behavioural  
614 genetics has shown that timing plays a critical role in regulating gene-environment  
615 interactions and, consequently, in determining developmental outcomes (Lenroot & Giedd,  
616 2011 cited in Levy & Eilam, 2013). In our study, the children with DS started to produce  
617 their first words on average 10 months later than their neuro-typical counterparts. This may  
618 be a small difference in relation to the human life span, but this initial delay can, over time,  
619 lead to a significant deviation from typical expressive language, and possibly to what may  
620 look like an isolated “domain specific” impairment in expressive language later in  
621 development. Future research should focus on considering how small variations in the early  
622 stages of development can develop into domains of relative strengths and weaknesses  
623 (Karmiloff-Smith, 1998).

624           From a clinical point of view, studying the developing phenotypes from its earliest  
625 origins is particularly relevant when considering early interventions, as there may be critical  
626 windows of opportunity in the early stages of development that could be targeted before they

627 become areas of significant weakness, or areas of early strength may be identified through  
628 which targeted intervention can be channelled. Currently there are few published intervention  
629 studies for young infants with Down syndrome. Having in depth knowledge of the how  
630 language progresses in the first 2-3 years of life can open opportunities for clinicians to  
631 develop ways of optimising language outcomes from the earliest stages of development.

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