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Specificity of Cognitive and Socio-Emotional Developmental Profiles in Children with Down Syndrome Presenting Diagnosis of Autism Spectrum Disorder

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Abstract

Background. The study compared the cognitive and socio-emotional development of children with Down Syndrome presenting diagnosis of Autism Spectrum Disorder (DSD-ASD), children with Down Syndrome (DS) and children with ASD. Participants. Fifteen children by group with mean different chronological ages (CA) and developmental quotients (DQ) were strictly matched on global developmental ages (respectively GDA : 18 months and 16 days, 19 months and 3 days and 18 months and 17 days). Methods. Cognitive and socio-emotional development, developmental ages and quotients assessments were carried out using the Social Cognitive Evaluation Battery (SCEB). Results. DS-ASD children had similar mean cognitive and socio-emotional developmental levels that both other groups but a significantly lower developmental level than in DS children in only one cognitive domain, Object relation schemata, and conversely, obtained a significantly higher level than DS children in only one socio-emotional domain, Affective Relations. Moreover, there was no significant difference for all cognitive and socio-emotional domains and heterogeneity indices between DS-ASD children and children with ASD. Conclusions. Relative to both DS and ASD children, children with DS-ASD exhibited a few specific developmental delays that showed evidence of the necessity of very early and intensive interventions centered on these disabilities.

Keywords: Down syndrome; Autism spectrum disorder; Dual diagnosis; Cognitive and Socio-emotional Development profiles; Socio-emotional and Cognitive Evaluation Battery

Abbreviations: ASD: Autism Spectrum Disorder; DSD-ASD: Syndrome presenting diagnosis of Autism Spectrum Disorder; DS: Down Syndrome; CA: Chronological Ages; DQ: Developmental Quotients; SCEB: Social Cognitive Evaluation Battery; ID: Intellectual Disability

Introduction

Autism spectrum disorder (ASD) is characterized by difficulties in communication and social interaction, and the presence of restricted or repetitive behaviors, interests, or activities. Symptoms are obvious in early childhood and limit the daily functioning of the child [1]. The notion of an autism spectrum was introduced to reflect the wide variability in the expression of symptoms from one individual to another. Thus, for each criterion, the degree of severity of the disorders presented

by the child is specified depending on the level of help he/she requires. In addition, the DSM-5 states that ASD diagnosis should not only specify the intellectual functioning and language level of the child, but also the presence of comorbidity and the association with a medical, environmental, or genetic illness such as Down's syndrome (DS).

Epidemiological studies report that 5 to 10% of people with DS have an additional diagnosis of autism [2-8], and 5 to 40% one of ASD [3,4,7,8]. Moreover, recent evidence shows that 16% to 18% of children with DS also meet diagnostic criteria for ASD [9]. Prevalence of ASD is considerably higher in people with DS than in the general population, in which 1 child in 150 is concerned [10,11]. Moreover, as this dual disorder remains poorly identified, its prevalence may be underestimated. Oxelgren, et al. [12] noted that 17 (13 males, 4 females) of the 41 children (41%) with DS in their population-based cohort met criteria for ASD, among which 53% also met the criteria for ADHD, the later exhibiting the most severe intellectual difficulties.

According to etiological research, there are two reasons for this higher prevalence of ASD in genetic syndromes: shared genetic etiologies [13], and fewer chances of cognitive compensation of the autistic symptomatology, independently inherited due to intellectual disability [2,6].

Down's syndrome is a genetic disease associated with an intellectual disability of variable severity degrees. Impaired abilities in social relationships and communication, as well as behavioral problems, are often observed. Thus, ASD and DS behavioral symptoms overlap, particularly when there is a severe intellectual disability, making the ASD diagnosis particularly difficult in a child with DS [4,5,11]. Nevertheless, Starr, et al. [14] showed evidence of ASD in children with DS at a severely/ profoundly retarded level of cognitive functioning. Early identification of ASD within the population of children with DS also seems compromised by professionals' lack of knowledge about the characteristics of children with developmental disorders, such as ASD and DS [2,11,15,16]. As a result, behavioral and developmental features tend to be incorrectly attributed to DS, without considering the possibility of an additional diagnosis of ASD. Being able to diagnose ASD in a child with DS requires good knowledge of the child's development and functioning. It also requires knowing how to use the assessment tools for these two developmental disorders [17], notably those for ASD, which enable the identification of individuals with DS and atypical [18].

As recognized by international classifications, intellectual disability and genetic syndromes may complicate the diagnosis of an associated ASD. Nevertheless, studies show that, even in people with a low developmental level, the diagnostic tools for autism are sensitive enough to identify whether there is an associated ASD [6,10,19,20]. The literature and clinical observations show that children with ASD present developmental and behavioral profiles that are distinct from those of children with an intellectual disability, even a severe one. In fact, intellectual disability (IQ less than 70) affects more than half of children with autism [21,22]. However, more recently, Christensen et al. [23] showed that the percentage of children with an intellectual disability (ID) varied widely between geographic areas, ranging from 20% to 50%. Brown et al. [24] indicated that about 40% of people with ASD are likely to present intellectual disability.

These children with ASD and intellectual disability present specific alterations in their communication and social interactions and show heterogeneities in verbal/non-verbal [25,26], socio-emotional and cognitive developmental profiles [27-29]. Children with an intellectual disability present an overall delay in acquisitions, reflected in the different areas of development [30], whereas children with DS follow a similar sequence of acquisitions to that of typical children, as well as a similar structuring of cognitive and communication skills, although their cognitive and communication development is delayed [31,32].

Studies have revealed clinical signs that can be used to identify the specific behaviors of children with a dual diagnosis of DS and ASD, even if their form varies from one individual to another [8,33], and highlighted a behavioral profile of children jointly affected by DS and ASD. When matched on the severity of their autistic symptomatology, individuals with a dual diagnosis and those with ASD present a similar behavioral profile, which differs from that of individuals with DS without an associated disorder [7]. This profile is characterized by more stereotyped behavior, repetitive language, overactivity and self-injury, but individuals with DS-ASD and DS appeared less withdrawn from their surroundings than those with ASD. Notably, as also recently noted by Moss et al. [34], children with DS display better sociability in all social contexts evaluated with familiar and unfamiliar adults (group, receiving interaction, initiating interaction, ongoing interaction), than children with other genetic syndromes and ASD. Thus, some characteristics bring together people with a dual diagnosis and those with DS, while distancing them from people with idiopathic ASD, notably social withdrawal and impulsivity [5]. In fact, Warner et al. [9] showed that in England and Wales, the profile of autism characteristics in children with DS-ASD was atypical compared with individuals with idiopathic ASD: they noted more emotional symptoms, conduct problems and hyperactivity. Moreover, Salehi et al. [35] compared a subgroup with DS-ASD to a group with ASD alone and found higher stereotype and lower irritability scores in the DS-ASD subgroup, on the Aberrant Behavior Checklist (ABC). Thus, children with DS-ASD might be considered both as a sub-group of the population of children with DS, as well as of children with typical ASD.

Despite growing literature over the last twenty years and the recognition by international classifications of the possibility of an additional diagnosis of ASD in people with DS, the issue has long been and still is neglected [36]. Some studies have described this dual pathology through behavioral symptoms but very few in developmental terms. Thus, the objective of this study was

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to identify the specificities of the cognitive and socio-emotional development of children jointly affected by DS and ASD, using a differential analysis of developmental profiles by comparing them with (i) children with DS, and (ii) children with ASD, assessed by means of the Socio-emotional and Cognitive Evaluation Battery (SCEB) [37].

The hypothesis was that children with a dual diagnosis would have a cognitive and socio-emotional developmental profile that differed from that of children with DS, and specially in socio-emotional domains, and was close to that of children with ASD in all domains.

Methods

Participants

The study was based on three groups of children with distinct diagnoses (Table 1): 15 children with a dual diagnosis of DS and ASD (mean age = 101 months; *SD* = 39,8; 3 girls and 12 boys; sex ratio = 0.25), and two other groups for comparison: 15 children with DS (mean age = 45 months; *SD* = 22,1; 2 girls and 13 boys ; sex ratio = 0.16) and 15 children with ASD (mean age = 59,7 months; *SD* = 27,5; 1 girl and 14 boys ; sex ratio = 0.07). The three groups had significantly different mean chronological ages ($F(2,42)=13.56$, $p<.0001$, Eta-squared = .39) (Table 1), but Scheffe post-hoc analysis showed that only both DS and ASD groups did not differ on this variable ($p=.43$) and that DS-ASD group differ from both the other groups (respectively $p<.0001$ with DS and $p=.003$ with ASD). All children with ASD or with DS-ASD received the diagnosis of autistic disorder according to the criteria of ICD-10 (F.84.0) and DSM-IV-TR and were later confirmed ASD based on DSM-5 criteria. Quantitative diagnostic assessment was also confirmed for all children by administering the Childhood Autism Rating Scale (CARS) [38,39], rated by authors who had carried out the developmental assessments of the children included in the study. Intellectual disability diagnosis was established from developmental assessments with the Socio-emotional and Cognitive Evaluation Battery, SCEB (Adrien, 2007), for all the children as described above. Diagnoses were performed by pedopsychiatrists, psychologists and geneticists experienced with ASD, DS and other neurodevelopmental disorders.

CARS scores of children were presented in Table 1. The degree of severity of autistic symptomatology between both groups of DS-ASD and ASD children was not significantly different ($t(28)=0.23$, $p=.82$, Eta-squared = 0.002).

Table 1: Characteristics of the study participants.

N = 45	DS	DS-ASD	ASD	p
	(n=15)	(n=15)	(n=15)	
Mean chronological age (months; days)	45; 0	101; 3	59; 7	<.0001
Standard deviation	22.1	39.8	27.5	
Min – Max	21 – 87	59 – 178	24 – 103	
CARS score	NA	39	38.6	.82
Standard deviation	NA	3.5	5.6	
Min – Max	NA	33.5 – 46	32 – 51.5	
Note. a. ASD refers to Autism Spectrum Disorder, DS refers to Down syndrome.				

Procedure

Children were recruited in day-care and consultation centers, associations, and parent forums. Information sheets and consent forms were given to parents. The study conforms to the standards of the Declaration of Helsinki and the European Medicines Agency Guidelines for Good Practice. It was approved by the Ethics Committee of Paris Descartes University (CERES N° 2013-36) and the CNIL (French Data Protection Organization: Decision DR-2014-436).

Evaluation sessions were carried out either in the children's homes or in their day-care center, by three researchers trained to use the tools, and with expertise in the clinical assessment of children with atypical development (1st, 3rd and 6th authors). During these sessions, the parents or the professionals who knew the children well were interviewed and the diagnostic evaluation using the Childhood Autism Rating Scale (CARS-T) and the child's developmental assessment using the SCEB were carried out.

Material and developmental measures

The Socio-emotional and Cognitive Evaluation Battery (SCEB) is a developmental test, designed and validated for children with autism whose developmental level ranges from 4 to 24 months of age [27,40]. Used in clinical practice and research [28,41,42], it was developed based on existing assessment scales for the cognitive, communicative, and emotional development of young children, as well as of Piaget's sensory-motor development theory [43] and Fisher's integrative developmental theory [44]. According to the latter, children's cognitive and social abilities undergo an integrated development, because of children's actions on their physical environment and interactions with people close to them. These people may play the role of experts in facilitating and developing children's cognitive and socio-emotional learning. Using situations and activities attractive to children, the SCEB evaluates 16 cognitive (n = 7) and socio-emotional (n = 9) domains. The seven cognitive domains are Object Permanence

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(OP), Operational Causality (OC), Spatial Relations (SR), Means-End (ME), object-relation Schemata (SCH), Self-Image (SIM), and Symbolic Play (SP). The nine socio-emotional domains are Behavior Regulation (BR), Social Interaction (SI), Joint Attention (JA), Expressive Language (EL), Receptive Language (RL), Vocal Imitation (VI), Gestural Imitation (GI), Affective Relations (AR) and Emotional Expression (EE). The items of these domains are classified into 4 developmental levels corresponding to psychological developmental ages: Level 1 = 4-8 months, level 2 = 8-12 months, level 3 = 12-18 months, level 4 = 18-24 months. Each item was rated, either as grade 2 (= complete success), grade 1 (= emergence or relative success with a bit of help and a demonstration) or grade 0 (= failure despite some help and a demonstration). The developmental level reached by the child in a domain corresponds to a level in which at least one of the items among the higher level was graded 1. A developmental level score from 1 to 4 was determined for each of the 16 domains and this provided a developmental profile for each child. By convention and to analyze the data, children who do not reach level 1 in a domain are attributed level 0 for this domain. A mean global development level (mean of 16 domains levels), a mean cognitive developmental level (mean of 7 domains levels) and a mean socio-emotional developmental level (mean of 9 domains levels) can be calculated. Moreover, the child's global, cognitive, and socio-emotional development may be homogeneous (without gap between levels in domains) or heterogeneous (with gap between levels in domains) and this heterogeneity may be objectified by calculating the overall (OHI), cognitive (CHI) and socio-emotional (SHI) heterogeneity indices.

Scores of the SCEB presented all reliability and validity qualities, according to the usual psychometric criteria [29,37].

Developmental ages and quotients

Assessment using the SCEB provides developmental levels in 16 domains (level scores: -0-, 1, 2, 3 and 4), a total score corresponding to the sum of the 16 domains developmental levels scores, a developmental profile in all 16 domains and an overall mean level score [37]. The transformation of the overall mean level score into Developmental Age (DA) can be based on the following empirical argument for validity: There is a strong correlation between mean SCEB level scores and the chronological ages observed for a sample of typical children ($r(104)=.94$, $p<.001$) [45,46]. These observations allow determining developmental ages (DA) from the SCEB overall mean level scores, by simply rescaling these last SCEB scores (level 0 to 4) on a scale ranging from 0 to 24 months (ratio 6). The DAs, as defined with rescaling, are perfectly correlated.

Thus, Developmental Quotients are overall mean level scores SCEB (0 to 4) rescaled on 0 to 24 (months) (DA), divided by chronological age (CA) and multiplied by 100. The mean developmental ages and quotients of the three groups of children are presented in Table 2.

Table 2: Mean developmental ages (AD in months) and quotients (DQ) of the study participants.

N = 45	DS	DS-ASD	ASD	*p*
	(n=15)	(n=15)	(n=15)	
Mean Developmental Ages	19.1	18.6	18.6	.89
Standard deviation	3.9	2.7	3.3	
Min – Max	11.1 - 24	13.1 - 23.3	12.4 - 24	
Mean Developmental Quotients	52	20.6	38.5	<.0001
Standard deviation	18.8	7	19.7	
Min – Max	20.3 – 79.9	12.0 – 34.9	13.0 – 84.5	
Note. a. ASD refers to Autism Spectrum Disorder, DS refers to Down syndrome.				

The three groups had not significantly different mean developmental ages ($F(2,42)=0.12$, $p=.89$, Eta-squared = 0.002 (Table 2). Moreover, the three groups had significantly different mean developmental quotients ($F(2,42)=14.17$, $p<.0001$, Eta-squared = .40, but Scheffe post-hoc analysis showed that only both DS and ASD groups did not differ on this variable ($p=.087$) and that DS-ASD group differ from both the other groups (respectively $p<.001$ with DS and $p=.016$ with ASD) (Table 2).

Thus, we can note on one hand, that the three groups of children were matched on developmental ages and on the other hand that the degree of severity of children's intellectual disability was more important as their chronological ages were high.

Data analysis and processing

Analysis was carried out with Stata for Windows (version 14). A multiple regression analysis was conducted on the developmental levels and heterogeneity indices obtained with the SCEB scale, comparing DS-ASD children to DS children and DS-ASD children to ASD children, i.e., using dual diagnosed children as a reference group for both simple diagnostic groups. While children with DS-ASD were significantly older and more intellectually disabled than the other children, comparisons were adjusted on chronological age and developmental quotient, and by convention, also on gender.

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Results

Development levels

Table 3: Comparison of SCEB global, cognitive, and socio-emotional developmental levels of the 3 groups of children.

SCEB Levels	A DS +ASD	B DS		C ASD	
	(N=15) Mean (SD)	(N=15) Mean (SD)	A vs B adjusted p level[b]	(N=15) Mean (SD)	A vs C adjusted p level[b]
Global Level	3.1 (.43)	3.19 (.61)	.45	3.1 (.53)	.37
Cognitive Level	3.33 (.36)	3.3 (.61)	.81	3.35 (.51)	.38
Socio-emotional level	2.99 (.61)	3.12 (.68)	.55	2.9 (.62)	.78

Note. a. ASD refers to Autism Spectrum Disorder. DS refers to Down syndrome. b. Regression analysis adjusted on gender. chronological age. and global developmental quotient.

There were not significant differences between the three groups for all the mean global, cognitive, and socio-emotional developmental levels (Table 3).

Regression analyses run separately on each domain showed significant lower level in DS-ASD children than in DS children in only one cognitive domain, Object relation schemata (Sch) (Table 4). Conversely, DS-ASD children obtained a significantly higher level than DS children in only one socio-emotional domain, Affective Relations (AR). Moreover, there was no significant difference for all cognitive and socio-emotional domains between DS-ASD children and children with ASD (see Table 4).

Table 4: Comparison of the domains' developmental levels in the cognitive and socio-emotional areas of the three groups of children.

(N=15)		DS + ASD	DS		ASD	
		(N=15)	A vs B	(N=15)	A vs C	
SCEB Area	SCEB Domain	Mean (SD)	Mean (SD)	adjusted p level[b]	Mean (SD)	adjusted p level[b]
Cognitive	SI	3.2 (.65)	3.4 (.95)	.63	3.73 (.57)	.19
	SP	3.07 (.77)	3.4 (.88)	.35	2.8 (1.17)	.99
	SCh	3.2 (.54)	3.6 (.61)	.049	3.27 (.57)	.26
	OC	3.07 (.57)	3 (.82)	.66	3.4 (.95)	.12
	ME	3.53 (.5)	3.4 (.8)	.27	3.33 (.6)	.4
	SR	3.93 (.25)	3.73 (.68)	.14	3.87 (.5)	.64
	OP	3.4 (.71)	3.2 (.83)	.75	3.07 (.77)	.57
Socio emotional	BR	3.8 (.4)	3.67 (.47)	.75	3.47 (.62)	.29
	SI	2.93 (.85)	3.2 (.75)	.57	3.07 (.85)	.68
	JA	3.27 (.93)	3.33 (.79)	.54	2.93 (1.06)	.28
	EL	1.93 (1.24)	2.8 (1.11)	.37	2.47 (1.31)	.37
	RL	3.4 (.61)	3.53 (.72)	.67	3.2 (.65)	.37
	VI	1.47 (1.5)	2.07 (1.44)	.18	1.67 (1.19)	.28
	GI	3 (1.46)	3.2 (.98)	.34	2.27 (1.24)	.7
	AR	4 (0)	3.73 (.68)	.036	3.8 (.54)	.15
	EE	3.33 (.6)	3.53 (.81)	.51	3.27 (.85)	.67

Note. a. ASD refers to Autism Spectrum Disorder. DS refers to Down syndrome.
b. Regression analysis adjusted on gender. chronological age. and global developmental quotient.

Legend:

Cognitive domains: SI: Self-Image. SP: Symbolic Play. Sch: Object relation schemata. OC: Operational Causality. ME: Means-Ends. SR: Spatial Relations. OP: Object Permanence

Socio-emotional domains: BR: Behavior Regulation. SI: Social Interaction. JA: Joint Attention. EL: Expressive Language. RL: Receptive Language. VI: Vocal Imitation. GI: Gestural Imitation. AR: Affective Relation. EE: Emotional Expression

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Heterogeneity indices

No significant differences were found between the three groups for the overall (OHI), cognitive (CHI) and socio-emotional (SHI) heterogeneity indices (Table 5).

Table 5: Comparison of overall (OHI), cognitive (CHI) and socio-emotional (SHI) heterogeneity indices of the 3 groups of children.

	A	B		C	
	DS + ASD	DS		ASD	
	(N=15)	(N=15)	A vs B	(N=15)	A vs C
	Mean (SD)	Mean (SD)	adjusted p level[b]	Mean (SD)	adjusted p level[b]
CHI	6.29 (2.9)	12.10 (5.6)	.06	10.25 (3.6)	.13
SHI	4.93 (3.7)	5.75 (5.0)	.13	5.54 (3.8)	.1
OHI	6.62 (4.7)	9.92 (6.6)	.2	8.51 (5.6)	.36

Note. a. ASD refers to Autism Spectrum Disorder. DS refers to Down syndrome. b. Regression analysis adjusted on gender. chronological age. and global developmental quotient

Discussion

This study presents some limitations. At first, the number of children in each group was small and so it is not possible to generalize the obtained results. Moreover, although children with different diagnosis in this study had similar developmental ages, children with DS-ASD were older than both the other groups. This might be explained by the fact that, in France, additional diagnosis of ASD in children with DS was later than in children who have only DS. So, young children with DS-ASD are few and so recruitment of this specific group of children with dual diagnosis was so biased. Further studies will have to focus on children with DS-ASD and low chronological ages, and maybe grouping international samples.

This study demonstrates that, as previously highlighted by behavioral analysis [7], children with a dual diagnosis of Down Syndrome and ASD seem to have a specific cognitive and socio-emotional developmental profile, which however is very slightly different from that of children with Down Syndrome without ASD, but quite like that of children with ASD.

Comparison of mean global, cognitive, and socio-emotional developmental levels

First, we noted that mean global, cognitive, and socio-emotional developmental levels were similar in the three clinical groups of children. While children with dual diagnosis were older and more intellectually disabled than children with DS, the absence of statistically significant differences shown after adjusting to chronological ages and developmental quotients was explained by similar global developmental levels. Thus, in our study, children with dual diagnosis, as a group, were not more delayed in their global, cognitive, and socio-emotional development than children with DS and children with ASD, even though the association of ASD as a comorbidity. And the same degree of severity of autistic symptomatology in DS-ASD and ASD children might explain the absence of differences in their mean global, cognitive, and socio-emotional developmental levels, confirming the point of view that social and communication deficits autism contributed to global developmental delay and intellectual disability [47].

Comparisons of cognitive and socio-emotional domains developmental levels

Moreover, the mean developmental levels of some cognitive and socio-emotional abilities in children with DS-ASD were not statistically different from those of other groups, although they were slightly lower, but except for two domains.

Cognitive area

When comparing all cognitive abilities developmental levels, there are no statistically significant differences except for object-relation Schemata (SCh), for which mean developmental level (=3,2) were slightly lower in children with a dual diagnosis than in children with DS (=3,6). This result show evidence that skills to coordinate and combinate several sensori-motor and social actions with objects and to produce substitutive play was less developed in older children with dual diagnosis. The greater maturation and a greater life experience of children with dual diagnosis attested by their higher chronological age did not imply a better or similar ability to produce coordinated sensori-motor and social and substitutive play actions than in children with DS. Thus, difficulties with fine motor skills due to low tone and/or hypermobility in hands, wrist or elbows known to be present in children with DS might be accentuated by the association of ASD in whom motor skills were also disturbed and delayed [49,50] and would persist despite longer interventions. Moreover, although mean developmental levels of Symbolic Play were not significantly different between both these groups, they were lower in children with DS-ASD (respectively 3,07 and 3,4). Moreover, while children with ASD was the lowest (2,8) in this domain, that was previously noted in children with ASD and severe intellectual disability [51-53], we can suggest that symbolic play's developmental delay in children with DS-ASD was explained by their autistic psychopathology.

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Socio-emotional area

In Behavior Regulation, Social Interaction, Joint Attention, and Emotional Expression domains, children with DS-ASD had mean developmental levels which were slightly lower but not statistically different than those in children with DS. This might reflect a rather good potential in socio-affective adaptation to others, despite the presence of ASD [7]. Surprisingly, we note that mean developmental level in Affective Relation was significantly higher in children with DS-ASD than in DS and in ASD. The assessment of this skill on the SCEB is based upon specific models of psycho-affective development [54] and focuses on the development of the self/other organization [55]. In this study, children with DS-ASD presented less significant delays in affective relation skills (level 4= 18-24 months) than children with DS (level 3= 12-18 months). This finding confirms the ability among severely intellectually challenged children with dual diagnosis to establish affective relations towards other people by affirmative and socio-adaptive behaviors, and to demonstrate self-awareness, despite their more severe communicative inabilities in expressive language and vocal imitation domains. Moreover, this may be explained by the fact that children with DS-ASD who are older have much more experienced emotional relationships with other people than do younger children with DS and with ASD. This was confirmed by Dykens et al. [56] research who noted that older children group with DS performed higher on the socio-adaptive Vineland scales than did younger children, although there was no significant difference.

On the other hand, children with DS-ASD had lower developmental levels in Expressive Language and Vocal Imitation domains than children with DS although there was no significant difference. In fact, these abilities are known to be particularly developmentally disturbed in autism [57,58] as confirmed by the similar low developmental levels in these domains in individuals with ASD in our study. In addition, while expressive language and vocal imitations mean levels in DS-ASD children were low but not statistically different from children with ASD, these data showed evidence of a severe deficit of language and vocal imitation abilities and functioning in this neurodevelopmental condition such as DS and ASD. In fact, in these children, the expressive language and vocal imitation developmental levels corresponded to the 4 to 8 months period of age, i.e., prelinguistic phase (production and imitation of vocalizations, sounds and double syllables…) while in other children with DS alone and with ASD alone, these developmental levels corresponded to the 12-18 months period of age (production of first words and association of two words, increasing in lexical stock…).

These domains are also usually delayed in ASD, such as found by Bernard Paulais et al. [28] in an international sample of children with ASD, and in children with Rubinstein-Taybi syndrome, a rare genetic disease usually associated to an intellectual inability, such as shown evidence by Taupiac et al, [59] and Adrien et al. [42]. However, in this study, children with ASD had a significantly higher expressive language developmental level than children with DS-ASD, despite the latter being older and having benefited from language rehabilitation and stimulation (about 8 years old vs. about 5 years of old for the children with ASD). While these disabilities in Vocal Imitation and Expressive Language are also observed in children with DS without autistic disorder [32], and whose cerebral dysfunction was described by Menghini et al, [60], the greatest neurodevelopmental vulnerability of these two verbal communication abilities might be due to the association of the genetic syndrome DS and ASD. Although there is no specific study focused on ASD and DS educational strategies, it seems that these two linguistic and imitation social abilities might require very early exploration and intervention plan, especially as adults with DS and DS-ASD benefit greatly from interventions centered on social skills [61].

Comparisons between heterogeneity indices

They were not significant differences between heterogeneity indices which confirms the similarity of the three groups ' developmental profiles.

To summarize, diagnosing ASD in children presenting with DS is essential to be diagnosed and must be considered as the primary disorder to offer the child and his/her family interventions following the current French recommendations [62,63]. In fact, it was necessary to provide early screening and management for these children [16], who require early intervention with qualitative appropriate approaches and programs that respond to their specific needs and differs from what is offered to children with the same genetic syndrome without an associated disorder [9]. It was suggested that these interventions might be focused on cognitive functions such as using various sensori-motor and symbolic schemes, vocal imitation, and expressive language functions, and based on the potential of socio-emotional functions such as behavior regulation, social attention, joint attention, gestural imitation, affective relations, and emotional expression functions which were as well developed as in children with DS alone. Moreover, both severely intellectually disabled and affected by ASD subgroup of children should not be forgotten in future treatment studies [64].

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Conflict of interests

The last author is the author of the SCEB edited in the Pearson France-ECPA. The other authors have no conflict of interests to declare.

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Ethic Statement

The study conforms to the standards of the Declaration of Helsinki and the European Medicines Agency Guidelines for Good Practice. It was approved by the Ethics Committee of Paris Descartes University (CERES N° 2013-36) and the CNIL (French Data Protection Organization: Décision DR-2014-436).

Informed Consent Statement

Written informed consent was obtained from all individual participants or their guardians included in the study.

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