Autism in the 6-12 months of life: pre-linguistic vocal trajectories and repetitive movements as markers of autism

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UNIVERSITÀ DI PISA

IRCCS FONDAZIONE STELLA MARIS

Montpellier 2017, September 15

ELENA Workshop

Déterminants des évolutions dans l'autisme

Cohorte ELENA

Genetic, Brain and Behavioral Heterogeneity

Gene X Environment

Brain adaptation
Connectivity

Response to treatment

Response to treatment

PRODROMIC STATE

Antecedents
Way of onset
Possible canalization

FULL SYNDROME

Clinical expression
Neuropsychological assessment
Functional assessment
Comorbidity/specificators

RESIDUAL STATE

Assessment of Changes
Longitudinal evaluation

Disentangling neurodevelopmental heterogeneity

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Review

Early detection of autism spectrum disorders: From retrospective home video studies to prospective 'high risk' sibling studies

Valeria Costanzo[a,1], Natasha Chericoni[a,1], Filomena Alessandra Amendola[b], Laura Casula[b], Filippo Muratori[a], Maria Luisa Scattoni[c,*,2], Fabio Apicella[a,2]

Retrospective
- Parental reports
- Homemovies

Prospective
- Screening tools
- High risk samples
  - Younger Siblings
  - Preterm infants

Trained observers, blind to group membership (TD vs ASD vs ID), examine child's behaviors (through specific grids, over time and across different very naturalistic situations) as they appear in Videos recorded by parents before diagnosis.
More recently this research has collected and analyzed HMs using computerized software (such as The Observer XT Noldus), a methodology that has increased accuracy and reliability of these studies.

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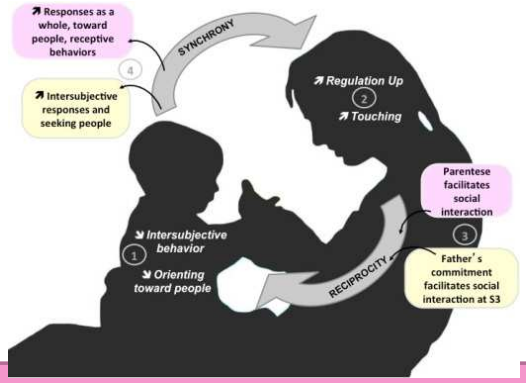
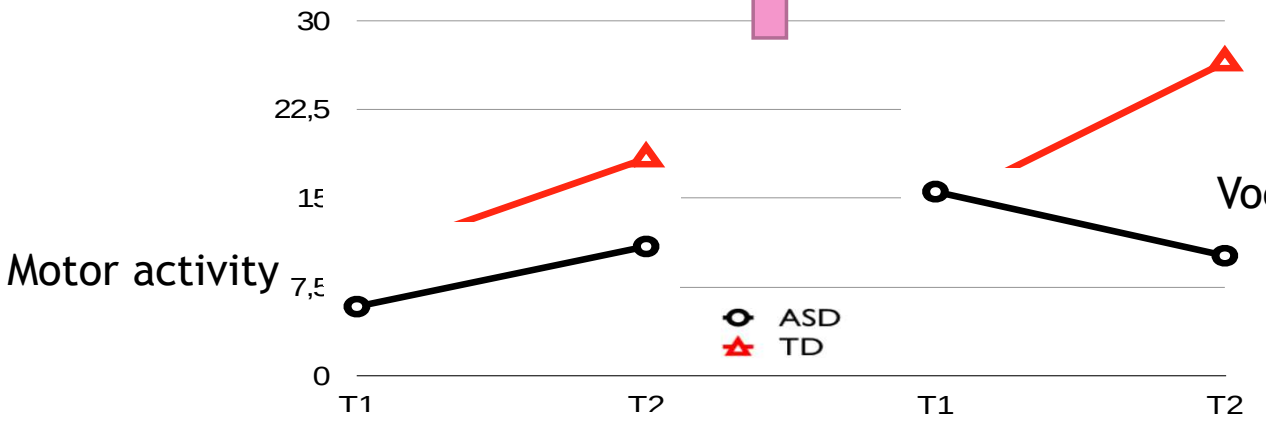
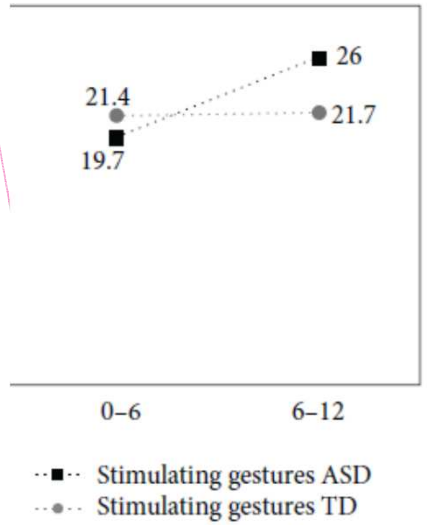
Research Article

Reciprocity in Interaction: A Window on the First Year of Life in Autism

Fabio Apicella,[1] Natasha Chericoni,[1] Valeria Costanzo,[1] Sara Baldini,[1] Lucia Billeci,[2] David Cohen,[3] and Filippo Muratori[1,4]







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ORIGINAL RESEARCH
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Bilateral Patterns of Repetitive Movements in 6- to 12-Month-Old Infants with Autism Spectrum Disorders

Giulia Purpura[1†], Valeria Costanzo[1†], Natasha Chericoni[1], Maria Puopolo[2], Maria Luisa Scattoni[3], Filippo Muratori[1,4] and Fabio Apicella[1]*

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ORIGINAL RESEARCH

Pre-linguistic Vocal Trajectories at 6–18 Months of Age As Early Markers of Autism

Natasha Chericoni[1], Daniela de Brito Wanderley[2], Valeria Costanzo[1], Andréa Diniz-Gonçalves[2], Mariluce Laitgel Gille[2], Erika Parlato[1], David Cohen[3,4], Fabio Apicella[1], Sara Calderoni[1,5] and Filippo Muratori[1,6]*

- Vocalization and babbling have been described as predictors of early language development and of its delay

- **Frequency of vocalization**, based on parent report, is predictive of language abilities in toddlers with ASD (Weismer, 2010)

- **Onset of canonical babbling** after 10 months has been shown to be a significant predictor of language delay (Oller, 1999)

Linguistic precursors

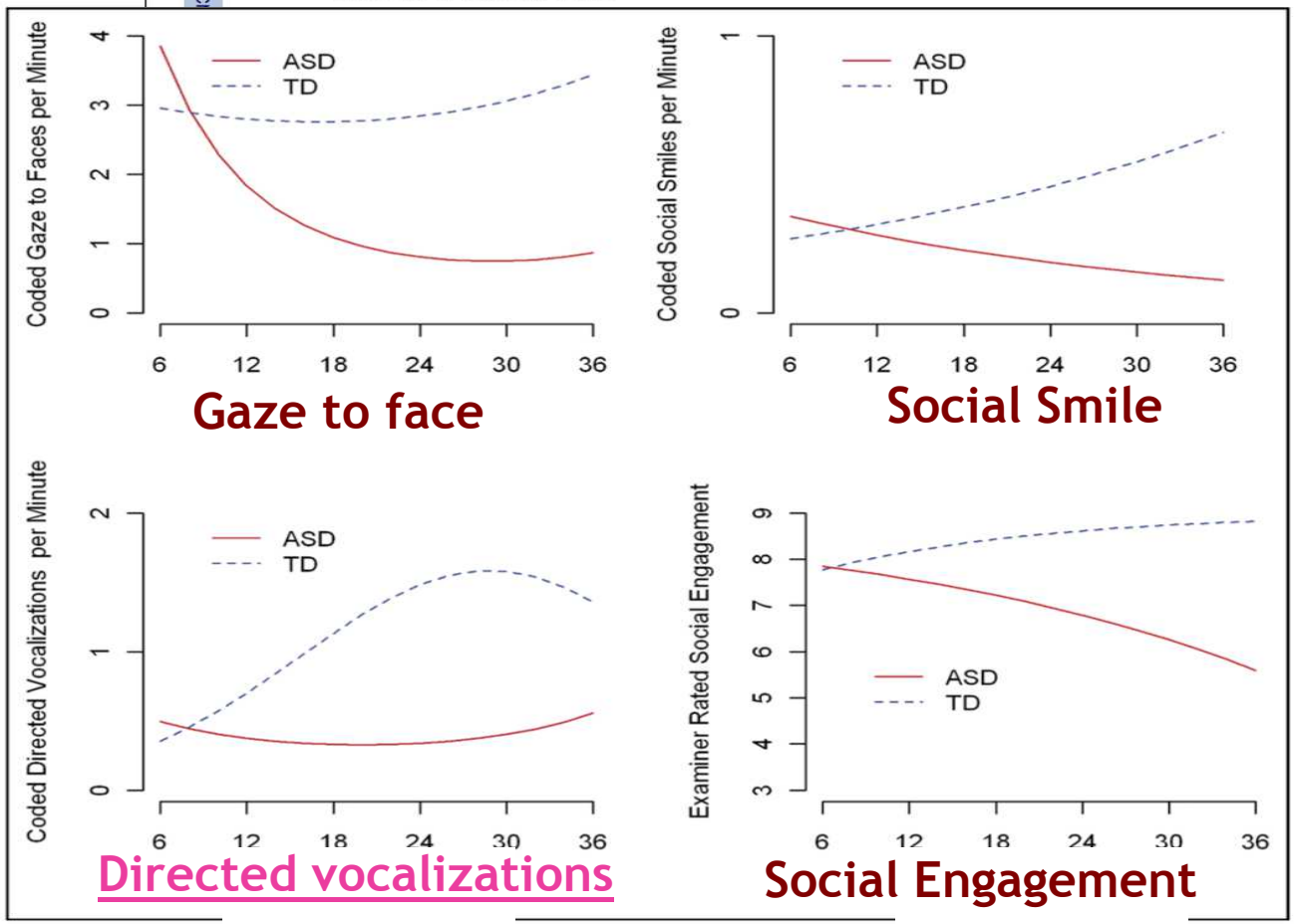
- From 6 months of life, ASD children show a gradual reduction of **directed vocalizations** and of other communication competences
- Trajectories start to be different between 6 and 12 months, and become significant at around **15 months** of life.

Published in final edited form as:
J Am Acad Child Adolesc Psychiatry. 2010 March ; 49(3): 256–66.e1-2.

A Prospective Study of the Emergence of Early Behavioral Signs of Autism

Dr. Sally Ozonoff, Ph.D.,
University of California–Davis

Ozonoff, 2010

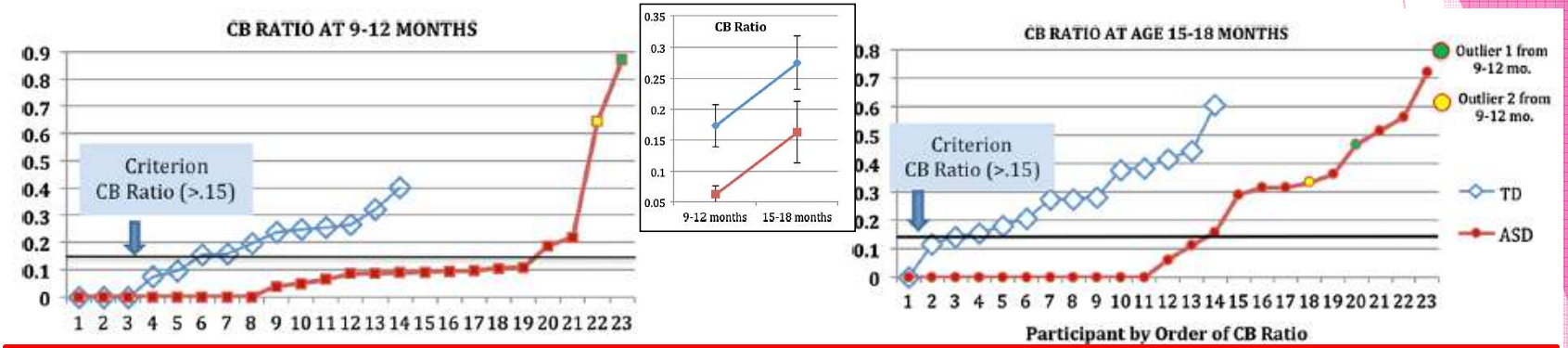


Vocal Patterns in Infants with Autism Spectrum Disorder: Canonical Babbling Status and Vocalization Frequency

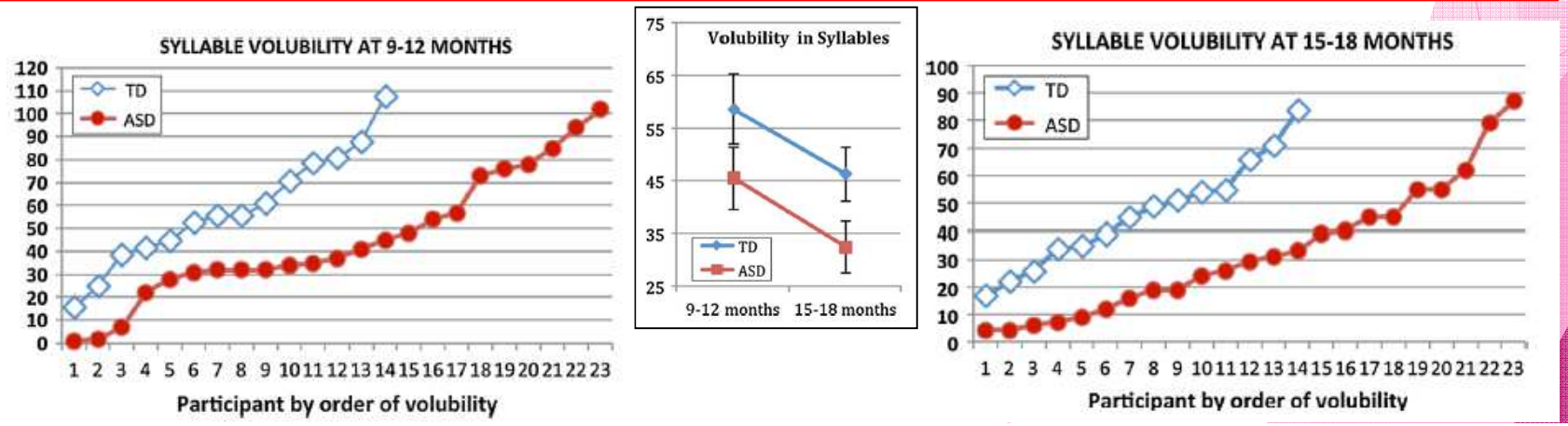
Elena Patten · Katie Belardi · Grace T. Baranek · Linda R. Watson · Jeffrey D. Labban · D. Kimbrough Oller

Home videos

Canonical babbling [ba] is comprised of a consonant-like sound and a vowel-like sound, with a rapid transition between them (Oller 1980, 2000).



Volubility: rate of vocalization measured in terms of frequency of syllable.



Vocalization Development in Toddlers With Autism Spectrum Disorder
Allison M Plumb & Amy M **Wetherby**
Journal of Speech, Language, and Hearing Research (2013)

- Examine the vocalizations of 125 children in the second year of life (**18 to 24 months**): 50 later diagnosed with ASD, 25 with developmental delays (DD), and 50 with TD.
- Measures of vocalizations were obtained through coding of video-recorded behaviors.
- **ASD:**
  - used a significantly **lower proportion of vocalizations with speech sounds**
  - used a significantly higher proportion of **atypical vocalizations** than children with TD.
  - used a significantly higher proportion of **distress vocalizations** than the TD and DD groups.
  - communicative vocalizations late in the second year were found to uniquely predict expressive language outcome at age 3 years above non-communicative vocalizations.

- **Conclusions** Type of vocalizations is a potential early indicator of ASD and **predictive of later language development**.

Vocalization Rate and Consonant Production in Toddlers at High and Low Risk for Autism
K Chenausky, C Nelson, **Helen Tager-Flusberg**
Journal of Speech, Language, and Hearing Research, April 2017

- Vocalization rate and consonant acquisition were investigated in speech samples at 12, 18, and 24 months from a prospective study of infant siblings of children with ASD. Three groups were compared: 18 toddlers at low risk for ASD, 18 high-risk siblings without ASD, and 10 high-risk siblings with ASD.
- **Results**
  - All groups' mean language scores were within the normal range.
  - At risk siblings with ASD showed consistently **lower vocalization rate**;
  - vocalization rate did not predict number of different consonants at 12 months for at risk siblings with ASD.
  - HRA−, not HRA+, toddlers had the smallest number of different consonants; Consonant-acquisition trajectories differed between groups, with HRA− showing the greatest increase from 12 to 18 months.
  - Lower vocalization rate was not associated with reduced number of different **consonants** in these toddlers.

Vocalizations

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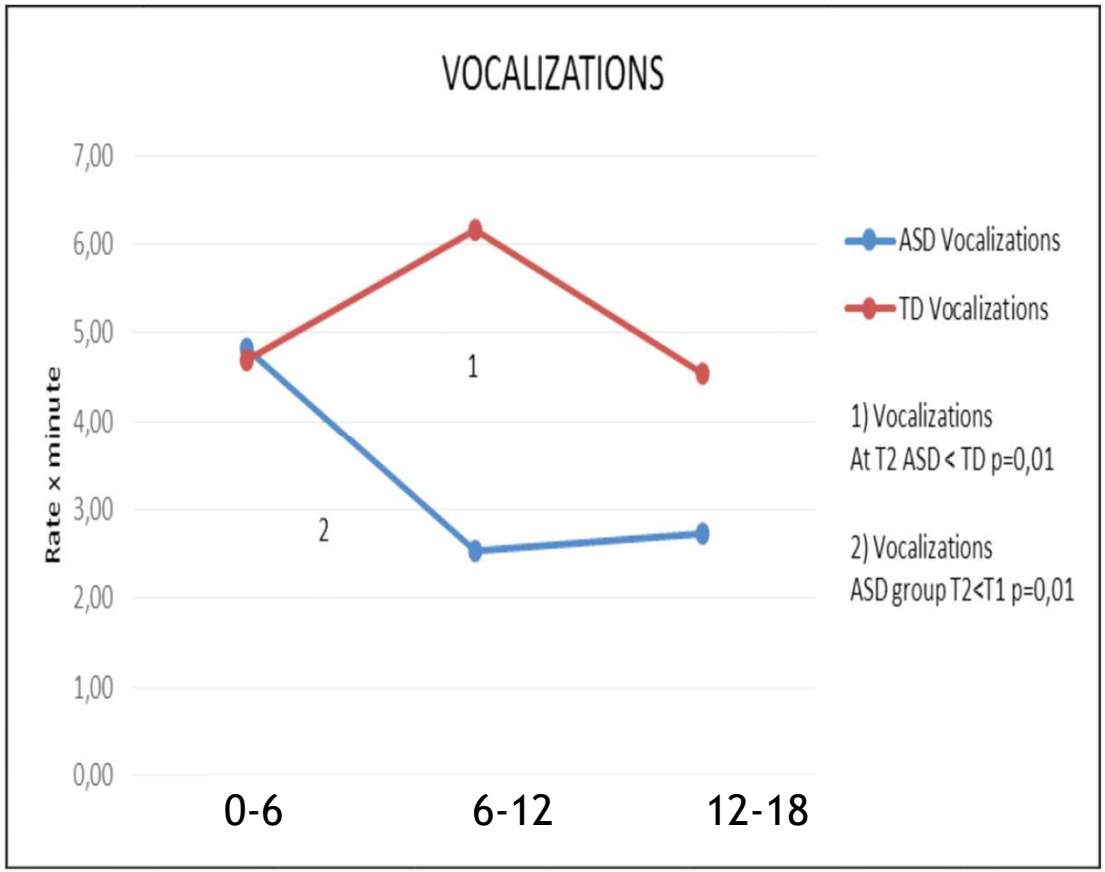

Similar rate of vocalizations characterize ASD and TD at 0-6 months.

During the 6-12 month period, vocal trajectories of ASD and TD infants diverge due to a significant sudden drop of vocalizations in ASD.

Frequency of vocalizations becomes significantly lower in ASD compared to TD.

Babbling

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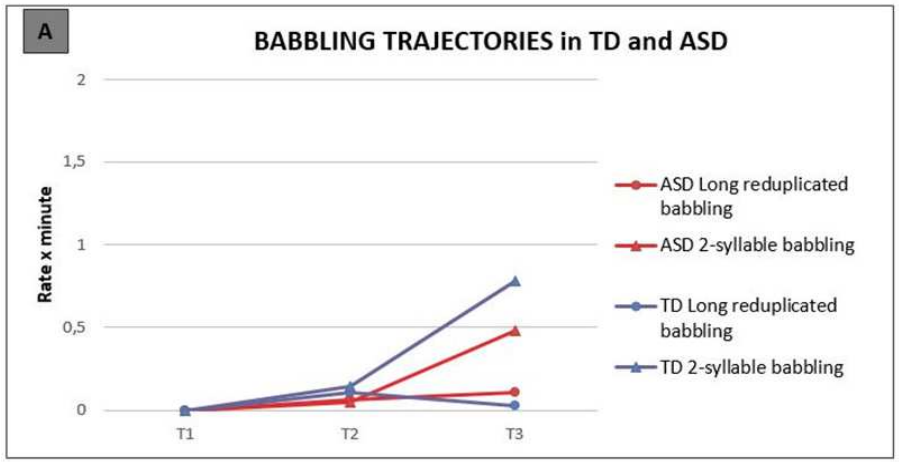
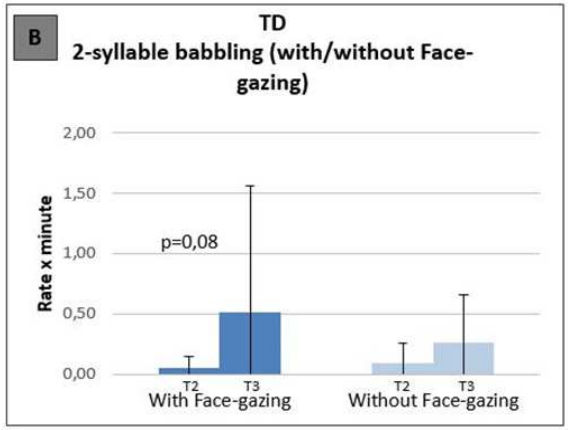
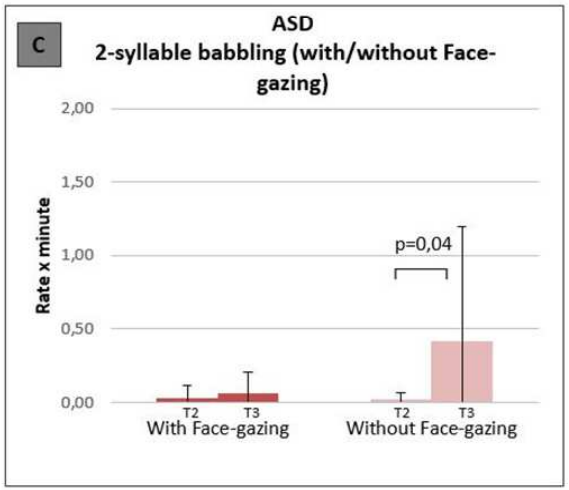
ASD toddlers do not show differences compared to TD toddlers, in terms of frequency (A), however a notable difference emerges in quality of babbling: ASD children show an increase of babbling not accompanied by face gazing, indicating a low interpersonal value of babblings (B,C).

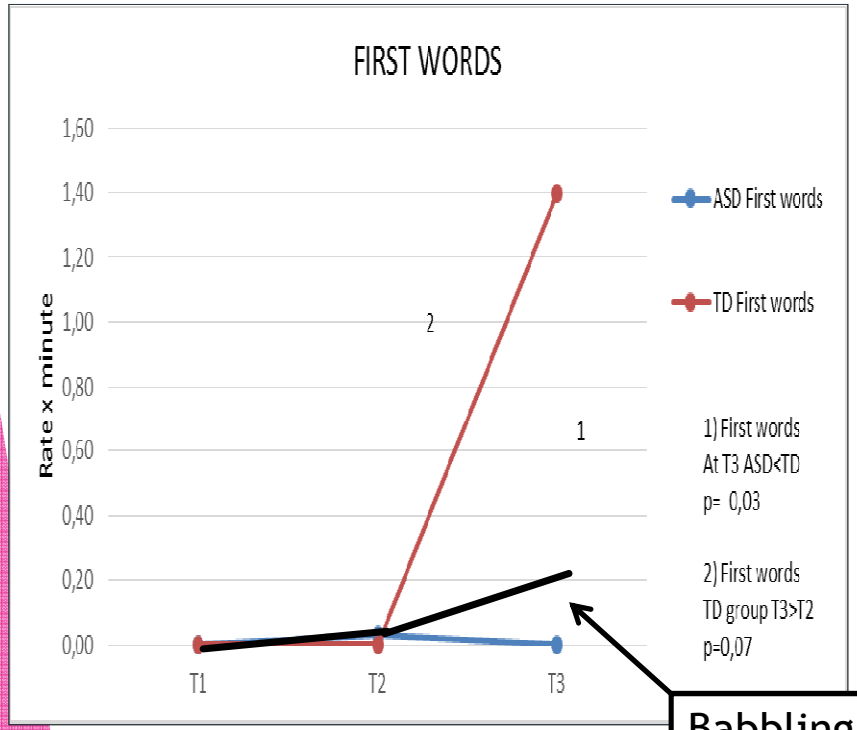
Social vs nonsocial babbling







First words



1) Contrary to ASD, TD toddlers show an increase in word production at 12-18 months.

2) At 18 months, word production is significantly lower in ASD compared to TD.

3) In ASD language remains for a longer period at the babbling level, with a clear prevalence of non social babbling compared to the socially directed babbling in TD.

Babbling in ASD (mainly nonsocial)

Language Differences at 12 Months in Infants Who Develop Autism Spectrum Disorder

DeWayne C. Lazenby[1,9] · Georgios D. Sideridis[1] · Noelle Huntington[1] · Matthew Prante[2,11] · Philip S. Dale[3] · Suzanne Curtin[4] · Lisa Henkel[4] · Jana M. Iverson[5] · Leslie Carver[6] · Karen Dobkins[6] · Natacha Akshoomoff[6] · Daina Tagavi[7,10] · Charles A. Nelson III[1,8] · Helen Tager-Flusberg[7]

Receptive and expressive language at 12 months

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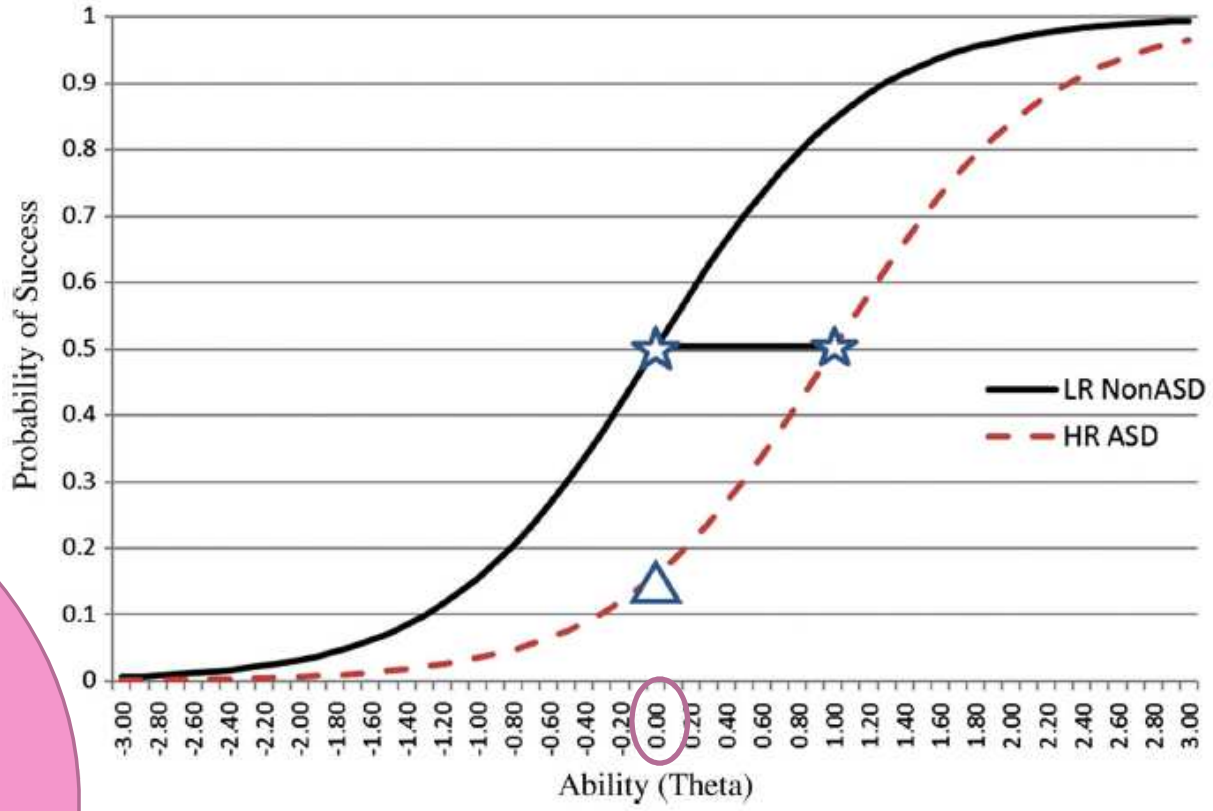
Lower receptive and expressive language scores in infants who later were diagnosed with ASD.

Table 2 Comparisons between groups on language measures

Score	High-risk		Low-risk
	ASD (N = 43)	Non-ASD (N = 170)	Non-ASD (N = 133)
	Mean (SD)	Mean (SD)	Mean (SD)
	Range	Range	Range
	N	N	N
Mullen receptive language	$37.61_{b,c}$	43.57_{a}	46.78_{a}
	(8.66)	(10.23)	(9.99)
T-score	20–55	20–79	31–80
	39	148	99
Mullen expressive language	$41.26_{b,c}$	46.817_{a}	51.13_{a}
	(11.20)	(11.44)	(9.02)
T-score	20–78	20–78	33–76
	39	148	98
CDI single word understanding raw	$35.88_{b,c}$	50.54_{a}	69.19_{a}
	N = 32	N = 165	N = 126
CDI single word production raw	2.05	5.84	6.06
	N = 37	N = 155	N = 120

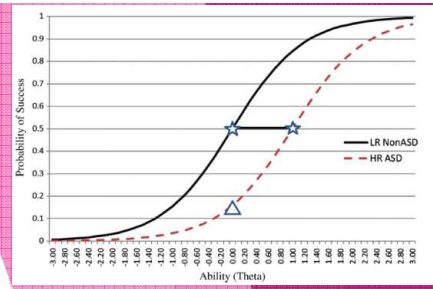
Probability of the word being used for infants of average language ability.

HR-ASD infants of average ability have a 15% probability of using a common word successfully(triangle), compared to 50% for the LR Non-ASD infants.

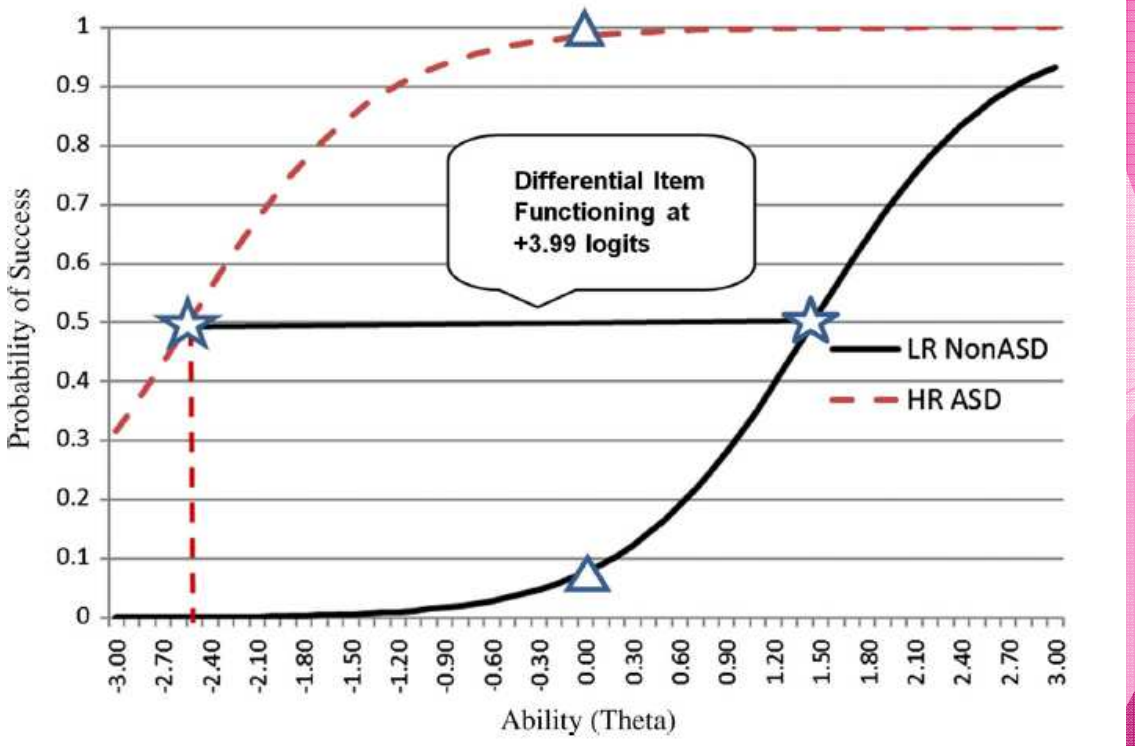


An **Ability** value of zero indicates average ability required for infants to have a 50% probability of producing or understanding the word; positive values indicate above average ability and negative values below average.

ASD had lower receptive and expressive language, but a higher degree of **statistically unexpected word** understanding and production.



Unexpected word (ie: "block") required a much lower level of ability for HR-ASD to be produced compared to the LR-Non-ASD (stars). At an average ability HR-ASD have a much higher probability of success (triangles)



Summarizing

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- ✓ Reduction of vocalizations
- ✓ Higher frequency of non-social babbling
- ✓ Fewer number of words

and more

- ✓ *Fewer consonants*
- ✓ *Atypical vocalizations*
- ✓ *Special skills for uncommon words*

2-3 ms

VIDEO 1

VIDEO2

7-8 ms

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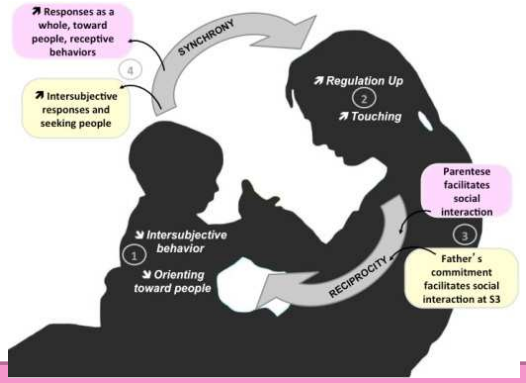

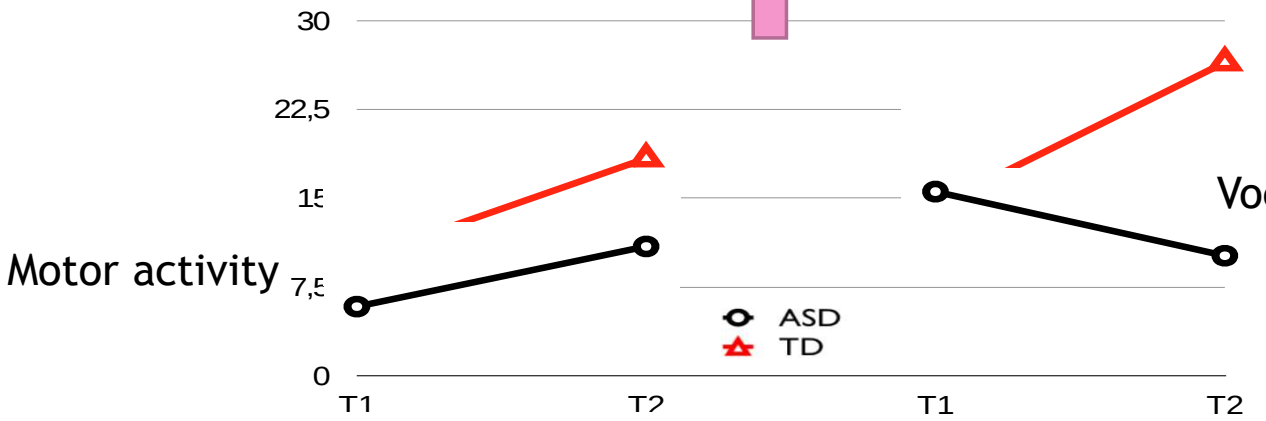

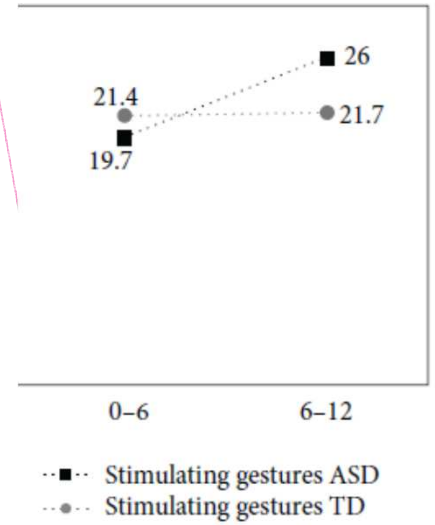

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ORIGINAL RESEARCH

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Motor disruption as a precursor of ASD

- **Motor disruption** (poor repertoire, hypomobility, less variability, asymmetry, odds movements) tends to be present in children with ASD early in life (Phagava, 2008).
- Abnormalities in body movements could be the first indicators of ASD (already during prenatal life?).
- Motor developmental step such as rolling over, sitting up, crawling and walking can be all abnormal and asymmetric (Teitelbaum, 1998).
- Motor coordination disorder is often described in ASD (Harris, 2017).

May we think that difficulties in maintaining social contact could be a downstream effect of motor disruption?

Motor Development

Asymmetry

Filippo MURATORI
Fabio APICELLA

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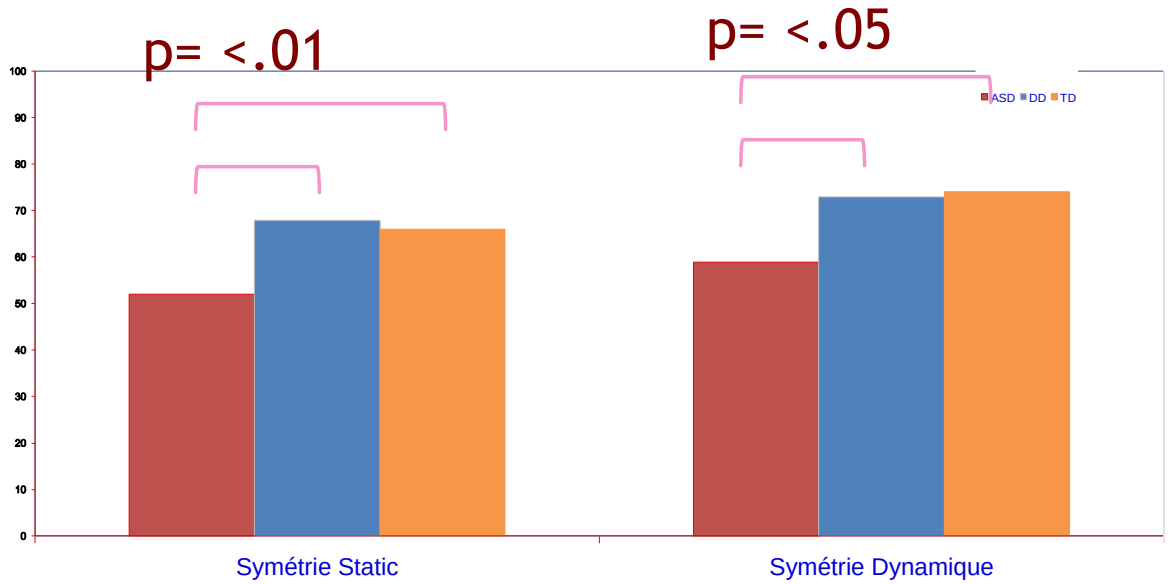
ASYMMETRY IN LYING POSITION



ELSEVIER

Brain & Development 31 (2009) 131–138

BRAIN & DEVELOPMENT
Official Journal of the Japanese Society of Child Neurology

www.elsevier.com/locate/braindev

Original article

An exploration of symmetry in early autism spectrum disorders: Analysis of lying

Gianluca Esposito [a,*], Paola Venuti [a], Sandra Maestro [b], Filippo Muratori [b]

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Motor Development

Asymmetry

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ASYMMETRY IN SITTING POSITION

VIDEO

Motor Development

Asymmetry

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ASYMMETRY IN FIRST GAIT

	AD		DD		TD			
	M	SD	M	SD	M	SD	$F_{(2,54)}$	sig.
WOS	$52,24_a$	9,49	$47,14_b$	5,79	$37,92_c$	4,69	20,98	**
F-Axis (Foot movement)	$60,81_a$	10,55	$50,04_b$	8,39	$41,70_c$	6,38	24,64	**
(f1) heel-toe pattern	$52,12_a$	15,87	$33,36_b$	10,57	$22,93_c$	6,42	31,76	**
(f2) tip-toe pattern	$75,06_a$	17,11	$66,72_b$	8,63	$56,30_c$	9,80	10,69	*
(f3) out-toeing pattern	55,25	13,99	50,04	17,27	45,87	12,84	2,08	ns
A-Axis (Arms movement)	$57,34_a$	21,90	$48,26_b$	11,48	$34,19_c$	11,00	10,64	*
(a1) forearm parallel to the ground, pointing forwar	$69,14_a$	24,13	$62,55_b$	19,31	$52,12_c$	15,87	3,57	*
(a2) arms are not held in a symmetrical position	$72,43_a$	22,38	$50,04_b$	10,57	$35,44_c$	15,28	14,64	*
(a3) elbow is in an irregular position	$66,94_a$	29,96	$50,04_b$	22,01	$38,57_c$	15,54	7,32	*
(a4) forearms are held rigidly	$69,14_a$	26,97	$66,72_a$	21,15	$37,53_b$	29,95	8,26	*
(a5) Arm-and-hand flapping	$23,04_a$	9,57	$11,94_b$	12,02	$7,26_b$	10,31	11,31	*
M-Axis (General movements)	$38,57_a$	5,28	$43,11_b$	6,53	$37,88_a$	4,20	4,7	*
(m1) myopathic gait or "waddling walk"	70,89	14,19	65,38	12,69	70,89	10,48	1,05	ns
(m2) lack of opposition pattern	$22,93_a$	9,33	$37,53_b$	8,63	$20,85_a$	7,68	16,16	**
(m3) stereotyped general movement	21,89	4,66	26,41	9,54	21,89	4,66	2,75	ns
PPSW								
Static Asymmetry	$59,50_a$	15,15	$32,05_b$	1,85	$33,75_b$	5,46	46,84	**
Dinamic Asymmetry	$49,05_a$	14,64	$26,80_b$	5,53	$26,50_b$	8,96	27,68	**

- ✓ Motor abnormalities may constitute a robust endophenotype for ASD (Esposito, 2010)
- ✓ The very early atypical development of motricity could results in difficulties in becoming intentional and in lesser use of motricity to be attuned to the intention of the other...
- ✓ …And make the child vulnerable to repetitive and stereotyped movements

Repetitive movements in TD

- Motor repetitiveness can be observed in TD when infants start to discover object's characteristics through repetitive movements that are necessary to train the action movements; this human behavior, "object banging", is displayed by TD infants during the second semester of life (Kahrs et al, 2013).

- Repetitive hand banging may be an opportunity for practicing rhythmically organized, tightly timed actions of the sort required for babbling (Iverson, 2010).

- Repetitiveness without object during the first year of life: TD show repetitive movements of arms, legs and hands, with a peak around 24 weeks of life, and a gradually reduction in frequency within the end of the first year (Thelen, 1979) .

- THUS, repetitive movements:
  - 1) seem to have an adaptive role in specific and limited temporal windows of development;
  - 2) it is supposed a continuum of such behaviors that extends from typical to atypical development (Leekam et al., 2007).

Repetitive movements in ASD

A corpus of researches examined the presence of repetitive movements at an early age, as likely related with the patterns of restricted and repetitive behavior in ASD (Loh, 2007; Morgan, 2008; Wolff, 2014; Elison, 2014).

These studies suggest that repetitive movements, from 12 months on, could be considered as a red flag for a ASD, even if it's not always clear if they are specific for this condition or represent a general risk factor.

VIDEO

A NEW HOME MOVIE STUDY

AIM

To point out if higher rate, duration and inventory of repetitive movements could differentiate infants with ASD from infants with DD or TD in the 6-12 months period of life when the disorder is in a prodromical phase of organization.

MATERIALS

Home Movies of infants later diagnosed with ASD, Intellectual Disability or TD

Focus on the 6-12 months age range

Sequences in which the entire body of the infants was clearly visible, randomly selected in order to obtain an equal distribution of situations (i.e bath time, feeding time, play time, etc) among groups.

A total of 8 minutes of videos *per* subject was selected; and a t-test was performed to verify if the selected material was comparable in terms of types of situations

A NEW HOME MOVIE STUDY

METHODS

- Behaviors coding was performed using a computer-based coding system: Observer XT 10.0 (Noldus, 2010).
- We developed a **Grid for coding Repetitive Movement Episodes** (RME) focusing on the entire movement repertoire and on specific body parts that are interested by repetitive movement in infants from six to twelve months
- **Definition of an Episode** of Repetitive Movement: the repetition for at least two times consecutively of an identical pattern of movement (flexion, extension, rotation, abduction, adduction or elevation in all possible directions) in a specific part of the body.
- Rate of episode per minute (FREQUENCY) and of percentage duration (DURATION) were obtained.
- ANOVA were computed in order to detect between groups differences. Due to the small sample, we set the level of significance at **$p<0.01$** in order to obtain the more powerful differences.

ITEM:	DESCRIPTION:	MODIFIER:
HEAD	Repeated pattern of movements of neck and head in all possible directions	
MOUTH	Repeated Oral movements without the presence of objects or body parts that approach or touch the mouth	
ARMS	Repeated pattern of movements of the arms starting from shoulder or elbow	(a) Bilateral (b) Unilateral
HANDS	Repeated pattern of movements of the hands starting from wrist, and without distal manipulation pattern with objects or body parts.	(a) Bilateral (b) Unilateral
HANDS WITH OBJECT	Repeated pattern of movements of the hands with distal manipulation pattern with objects or body parts	(a) Bilateral (b) Unilateral
HAND FINGERS	Repeated pattern of movements of the hand fingers starting from phalanx, and without distal manipulation pattern with objects or body parts.	(a) Bilateral (b) Unilateral
TRUNK	Repeated pattern of movements of trunk in all possible directions	
LEGS	Repeated pattern of movements of legs or/and feet starting from hip, knee or ankle, and without distal manipulation pattern with objects or body parts	(a) Bilateral (b) Unilateral

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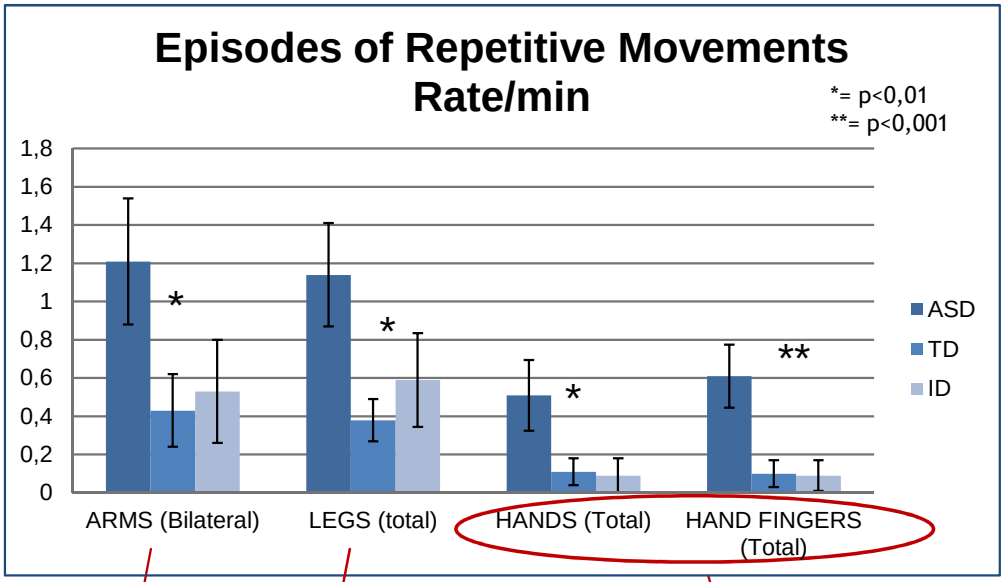
HANDS WITH OBJECT	Repeated pattern of movements of the hands with distal manipulation pattern with objects or body parts	(a) Bilateral (b) Unilateral
HAND FINGERS	Repeated pattern of movements of the hand fingers starting from phalanx, and without distal manipulation pattern with objects or body parts.	(a) Bilateral (b) Unilateral
TRUNK	Repeated pattern of movements of trunk in all possible directions	
LEGS	Repeated pattern of movements of legs or/and feet starting from hip, knee or ankle, and without distal manipulation pattern with objects or body parts	(a) Bilateral (b) Unilateral

RESULTS. 1 - FREQUENCIES



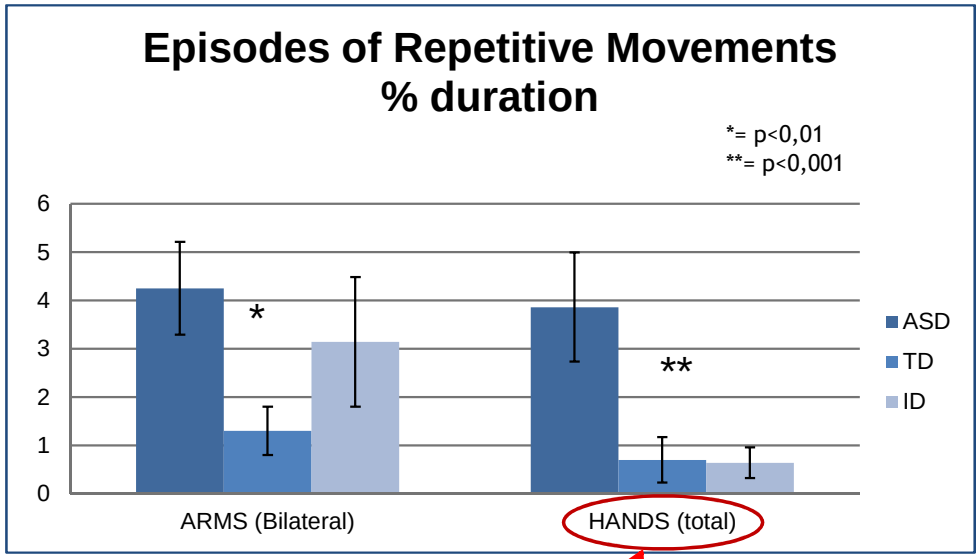
	Mean (SD)			Post-hoc test p-value		
	ASD (1)	TD (2)	ID (3)	1 vs 2	1 vs 3	2 vs 3
Arms bilateral	1,21 (0,66)	0,43 (0,39)	0,53 (0,55)	0,01*	n.s.	n.s.
Legs	1,14 (0,54)	0,38 (0,22)	0,59 (0,49)	0,002 *	n.s	n.s.

	Mean (SD)			Post-hoc test p-value		
	ASD (1)	TD (2)	ID (3)	1 vs 2	1 vs 3	2 vs 3
Hands	0,51 (0,37)	0,11 (0,15)	0,09 (0,23)	0,008 *	0,005 *	n.s.
Hand Fingers	0,61 (0,33)	0,10 (0,15)	0,09 (0,16)	0,000 **	0,000 **	n.s.

ARMS & LEGS: ASD show higher rate of repetitive movements than TD (not ID).

HANDS and FINGERS: Patterns of Repetitive Movements are specific to ASD

Results. 2 - DURATION



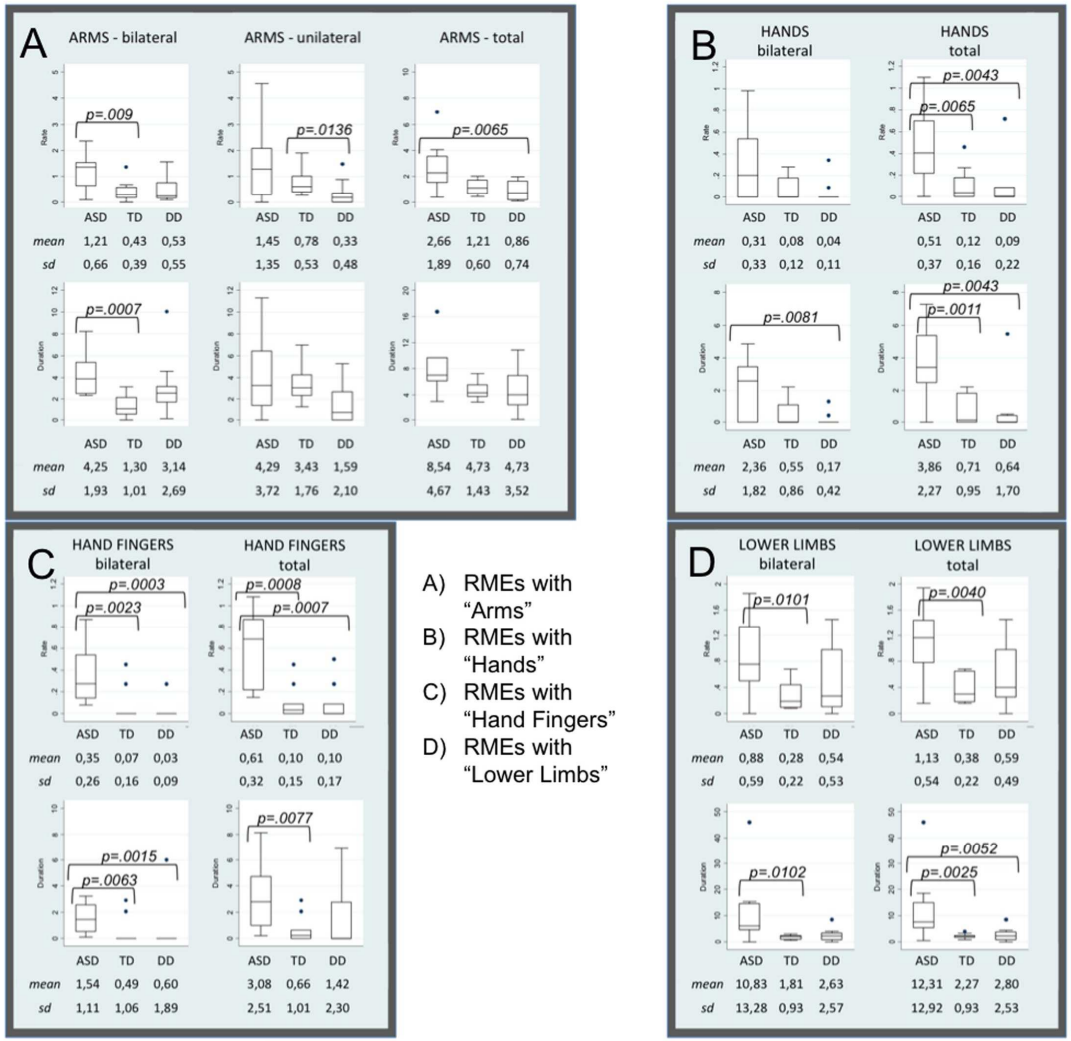
Specific for ASD

ASD spend more time in RMs with Arms and Hands

Differences were statistically significant only for **Bilateral** repetitive movements.

	Mean %d (SD)			Post-hoc t-tests p value		
	ASD (1)	TD (2)	ID (3)	1 vs 2	1 vs 3	2 vs 3
Arms (bilateral)	4,25 (1,93)	1,30 (1,01)	3,14 (2,69)	0,008 **	n.s.	n.s.
Hands (Total)	3,86 (2,27)	0,70 (0,95)	0,64 (1,70)	0,001 **	0,001 **	n.s.

No statistical differences between TD and ID.



A) RMEs with "Arms"
B) RMEs with "Hands"
C) RMEs with "Hand Fingers"
D) RMEs with "Lower Limbs"

ASD infants show higher rate and duration of RM in:

- ✓ **ARMS** bilateral (and total)
- ✓ **HANDS** bilateral (and total)
- ✓ **FINGERS** bilateral (and total)
- ✓ **LEGS** bilateral (and total)

Bilateral pattern of repetitive movements

- VIDEO 1

- VIDEO 2

- Presence of a specific pattern in infants with ASD as far as repetitive movements with **hands** and **hand fingers** during the second semester of life.

- Our results in the 6-12 month period could be considered in a longitudinal way as correlated to the earlier poor repertoire of General Movements (Phagava, 2008) and to the lack of variability in finalized movements described in the second year of life (Loh, 2007; Elison, 2014)

- We could suppose that, due to the fact that infants are frequently enganged in episodes of RMs with hands and fingers, they are impaired in their use to perform functional actions and in particular communicative gestures that usually emerge at this age range.

- The high frequency and duration of bilateral RMs with hands and fingers could suggest an atypical use of hand movements, that at this young age, as in a certain period of human phylogenetic development, typically start to be used to gesture and communicate with others (Corballis, 2002).

- Thus, the presence of RMs might reduce the possibility for a communicative use of hands.

- This hypothesis might be in line with evidence from literature that suggest a correlation between early motor dysfunction and language impairments.

The 6 to 12 month period of life: a special window to detect risk of autism?

- Sudden drop of vocalizations
- Prevalence of non social babbling
- Increase of repetitive finger movements.

AND

- Lack of response to name.
- (Lack of mirror stage (jubilatory reaction to the vision of its own image reflected in the mirror)?

VIDEO

- Thanks for your attention

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