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What do home videos tell us about early motor and socio-communicative behaviours in children with autistic features during the second year of life — An exploratory study

Michele Zappella [a], Christa Einspieler [b,*], Katrin D. Bartl-Pokorny [b], Magdalena Krieber [b], Mary Coleman [a], Sven Bölte [c,d], Peter B. Marschik [b,c]

[a] Foundation for Autism Research, Inc., New York, NY, USA
[b] Research Unit iDN — interdisciplinary Developmental Neuroscience, Institute of Physiology, Center for Physiological Medicine, Medical University of Graz, Graz, Austria
[c] Center of Neurodevelopmental Disorders (KIND), Department of Women's & Children's Health, Karolinska Institutet, Stockholm, Sweden
[d] Child and Adolescent Psychiatry, Stockholm County Council, Stockholm, Sweden

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ABSTRACT

Background: Little is known about the first half year of life of individuals later diagnosed with autism spectrum disorders (ASD). There is even a complete lack of observations on the first 6 months of life of individuals with transient autistic behaviours who improved in their socio-communicative functions in the pre-school age.
Aim: To compare early development of individuals with transient autistic behaviours and those later diagnosed with ASD.
Study design: Exploratory study; retrospective home video analysis.
Subjects: 18 males, videoed between birth and the age of 6 months (ten individuals later diagnosed with ASD; eight individuals who lost their autistic behaviours after the age of 3 and achieved age-adequate communicative abilities, albeit often accompanied by tics and attention deficit).
Method: The detailed video analysis focused on general movements (GMs), the concurrent motor repertoire, eye contact, responsive smiling, and pre-speech vocalisations.
Results: Abnormal GMs were observed more frequently in infants later diagnosed with ASD, whereas all but one infant with transient autistic behaviours had normal GMs ($p < 0.05$). Eye contact and responsive smiling were inconspicuous for all individuals. Cooing was not observable in six individuals across both groups.
Conclusions: GMs might be one of the markers which could assist the earlier identification of ASD. We recommend implementing the GM assessment in prospective studies on ASD.

1. Introduction

Delay in motor and/or speech-language development is often the first sign to raise parental and professional concerns, leading to further clinical examinations and assessments. In rare occasions, children may fail to progress after a period of relatively normal development or may even lose acquired functions (regression). A typical example is Rett syndrome (RTT), where regression usually occurs between 6 and 18 months of age, may last for several months, and is followed by a developmental plateau and a late deterioration stage [1]. Home video analysis of the prodromal period in RTT, previously considered inconspicuous, has demonstrated abnormalities in early spontaneous movements [2,3]. These findings contributed to our growing knowledge of the natural history of RTT, suggesting that the corresponding genetic mutations (*MECP2* in 95–97%; [1]) are operant before birth. The most frequently occurring pattern of early spontaneous movements is the so-called general movements (GMs). GMs are spontaneously generated by so-called central pattern generators that are most probably located in the brain stem and are modulated by more rostral parts of the brain [4,5]. Hadders-Algra proposed that the most important features of GMs, complexity and variation, are generated by the cortical subplate [6]. GMs can be observed from 9 weeks' gestation until 5 months after term, and their quality is considered one of the most reliable indicators for (dys)function of the developing nervous system [4–7].

Regression occurs in every third individual with autism spectrum disorder (ASD) [8]. Parental reports, retrospective video analyses, and prospective studies of infants at high risk revealed that individuals with ASD already had a number of more or less subtle atypicalities in their pre-regressional motor and socio-communicative behaviours (for a recent review, see [9]). Among the atypicalities observed as early as the first half year of life were postural asymmetries while lying [10,11], head lag during the pull-to-sit transition [12], fluctuating

* Corresponding author at: Institute of Physiology, Medical University of Graz, Harrachgasse 21, 8010 Graz, Austria. Tel.: +43 316 380 4266; fax: +43 316 380 9630.
E-mail address: christa.einspieler@medunigraz.at (C. Einspieler).

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muscle tone [13], oral-motor abnormalities [14], and abnormalities in GMs (for a recent review, see [15]).

Such early observations are lacking for a subgroup of toddlers with transient autistic behaviours during the second year of life. After several months or even years, they lose their autistic behaviours and eventually acquire age-adequate communicative and cognitive abilities, albeit often accompanied by tics and/or attention deficit [16–23]. To shed light on the early development of these individuals (i.e. from birth to 6 months), we collected family videos and compared early behaviours of individuals with such transient autistic behaviours with observations of infants later diagnosed with ASD. Our exploratory study focused on the following developmental areas: (a) GMs, (b) the concurrent motor repertoire, (c) eye contact and responsive smiling, and (d) pre-speech vocalisations.

2. Methods

2.1. Participants

Videos of 34 individuals (born in Italy between 2004 and 2010) who experienced autistic behaviours during their second year of life were donated by families for the assessment of movements, posture, and socio-communicative behaviour during the first 6 months of life. This material had been recorded by the families as part of their family archive without knowing that the child might eventually develop an adverse neurodevelopmental outcome. All parents reported the development within the first year of life as normal. For this study, we had a footage of 18 male individuals at hand that contained sufficient recordings performed during the first half a year of life. All boys had been born at term with an appropriate birth weight. Table 1 provides the age at the onset of autistic behaviours and the further neuropsychiatric development. The neuropsychiatric diagnoses were based on the assessment of the first author (who is a child psychiatrist) according to the criteria of *ICD-10*. Ten children (Cases 9–18) were diagnosed with ASD at an age between 3;6 and 7;1 (years;months). Cases 1–8 showed no autistic behaviours after age 3. Seven individuals with transient autistic behaviours were later diagnosed with Tourette syndrome (TS) (Cases 2–8); TS also occurred as a co-morbidity in four individuals later diagnosed with ASD (Cases 9, 10, 11, 18). Attention deficit hyperactivity disorder (ADHD) was found in five individuals with transient autistic behaviours (Cases 2, 4, 5, 7, 8), and occurred as a co-morbidity to ASD in Cases 9, 10, 11, 13, and 17. Case 1 showed no psychiatric disorder at his last examination at age 7;4 (Table 1).

The parents of the study cases gave their informed consent for participation in the study and publication of the results. The Institutional Review Board of the Medical University of Graz approved the method of retrospective video analyses.

2.2. Data analysis

Video sequences that were suitable for analysis (314 min in total; duration per infant for the 1- and 2-month analysis: median = 7 min [IQ: 6–11 min]; for the 3- and 4-month analysis: median = 4 min [IQ: 3–7 min]; and for the 5- and 6-month analysis: median = 6 min [IQ: 4–11 min]) included the following settings: the infant in supine and prone position, held semi-upright by the caregiver or in a relaxing chair, sitting with or without support, feeding, and bathing (Table 2). The videos were analysed by three scorers (C.E., K.D.B., P.B.M.) who did not know the further development of the infants.

Apart from age-specific swiping and wiggling–oscillating arm movements, kicking, movements towards the midline, and antigravity movements [24], we assessed the age-specific GMs. GMs have a writhing character during the first 2 months after term, and a fidgety character thereafter. Writhing GMs comprise the entire body and manifest themselves in a variable sequence of arm, leg, neck, and trunk movements. They come and go gradually, varying in intensity and speed. Rotations and frequent slight variations of the direction in motion make them appear complex and smooth [4]. Abnormal writhing movements are classified as (a) poor repertoire GMs, whereby the sequence of movement components is monotonous and the amplitude, speed, and intensity lack the normal variability; or (b) cramped-synchronised GMs which lack the usual smoothness and fluent character, appearing rigid as the limb and trunk muscles contract almost simultaneously and relax almost simultaneously [4]. Fidgety GMs (observable from 3 to 5 months and sometimes even a few weeks longer) are small movements of the neck, trunk, and limbs in all directions and of variable acceleration.

Table 1
Age and examination at the onset of autistic behaviours, and further neuropsychiatric development of our sample (n = 18).

Case[a]	Age at the onset of autistic behaviours in months	Examination at the age of onset	Age at the last examination in years;months	ASD according to the last clinical examination	Cognitive status at the last clinical examination[b]	Tourette syndrome	ADHD
1	18	Clin eval[c]	7;4	−, CARS = 15	Normal	−	−
2	18	ABC = 47	9;7	−, CARS = 18	Normal	+	+
3	18	ADOS = 8	7;0	−, CARS = 15.5	Normal	+(f)	−
4	18	CARS = 34	4;9	−, CARS = 25	−2 SD	+	+
5	18	Clin eval[c]	9;0	−, CARS = 15	−1 SD	+	+
6	18	Clin eval[c]	6;9	−, ABC = 3	−1 SD	+	−
7	20	CARS = 33	5;9	−, CARS = 16	Normal	+(f)	+
8	24	ADOS = 20	10;0	−, CARS = 17	Normal	+	+
18	14	Clin eval[c]	4;6	+, ASD[c]	−1 SD	+	−
10	15	CARS = 36	7;1	+, ASD[c]	−1 SD	+(f)	+
12	18	CARS = 44.5	6;4	+, ADOS = 19	Normal	−	−
9	18	ADOS = 10	7;1	+, HFA	Normal	+	+
14	18	ADOS = 20	3;10	+, ASD[c]	Not testable	−	−
15	20	Clin eval[c]	3;10	+, CARS = 41	−1 SD	−	−
11	12–24[d]	CARS = 37	7;0	+, ABC = 48	−2 SD	+(f)	+
13	12–24[d]	Clin eval[c]	4;3	+, ASD[c]	Normal	−	+
16	12–24[d]	Clin eval[c]	3;6	+, ASD[c]	−1 SD	−	−
17	12–24[d]	CARS = 32.5	5;10	+, ADOS = 18	Not testable	−	+

Key: ADHD = attention deficit hyperactivity disorder; ABC = Autistic Behaviour Checklist; ADOS = Autistic Diagnostic Observation Schedule, subcategory communication and interaction (cut-off value = 6); ASD = autism spectrum disorders; CARS = Childhood Autism Rating Scale; HFA = high-functioning autism; SD = standard deviation; + = present; − = absent; (f) = familial.

[a] Cases are rank-ordered within each group according to the age at the onset of autistic behaviours.

[b] According to the Wechsler Intelligence Scale for Children (WISC-III), Leiter Scale, or Wechsler Preschool and Primary Scale of Intelligence (WPPSI).

[c] Clinical evaluation according to the *ICD-10* criteria for autism spectrum disorders.

[d] During the second year of life, no further specification.