

## The General Movements in children with Down syndrome

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### Abstract

*Objective:* Aim of our study was to describe the character of General Movements (GMs) in children with Down Syndrome (DS).

*Material and methods:* GMs of 23 children with DS and of 30 healthy full-term infants were assessed from birth to 6th month corrected age. A qualitative and a semi-quantitative evaluation of GMs were achieved for each child. Data were graphically displayed to obtain growth curves of motor optimality scores.

*Results:* GMs in children with DS are characterised by low—low/moderate speed, large—large/moderate amplitude, partially creating impression of fluency, smoothness and complexity, abrupt beginning and end, few other concurrent gross movements. During the 6 months, all children showed an improvement of qualitative and semi-quantitative evaluation, but it was possible to observe great heterogeneity among children in the evolutionary course. GMs evaluation of children with no known motor problems was normal, showing only slight and transient abnormalities at first months.

*Conclusion:* GMs character of children with DS could be related to central nervous system and peripheral abnormalities characterizing this syndrome. The evaluation of GMs in children with DS could be an early marker of motor impairment and help in early management decisions making.

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## 1. Introduction

Down syndrome (DS), long been recognized as one of the most important causes of disability, is characterized by delays in development of motor function (as a result of associated impairments including muscle hypotonia, joint hyperextensibility, delayed acquisition of postural control, poor balance) and mental retardation [1].

Many brain alterations in children with DS have been described: reduced weight, decreased number and depth of sulci in the cerebral cortex, reduced number of specific populations of neurons, alterations of the cortical laminations, reduced dendritic ramifications, diminished synaptic formations, as well as functional abnormalities in membrane properties [2].

Motor development of children with DS has been studied by many authors primarily by recording the mean age of achievement of motor milestones [3–6]. Chen and Wolley [3], administering the Denver Developmental Screening Test in children with DS, observed that the developmental achievement quotient for the gross motor domain decreases in older children compared to younger; however, the sequence of items passed by children with DS was similar to the sequence of children with no known motor problems.

Russell et al. [7–9] demonstrated a good validity of the gross motor function measure (GMFM) for the assessment of children with DS, and created gross motor function growth curves of children with DS between the ages of 1 month and 6 years.

The aim of our study is to describe the character of General Movements (GMs) in children with DS and to compare it with a control group of healthy full-term infants. Spontaneous motor activity was studied with the Prechtl method, based on the video recording of GMs and the qualitative and semi-quantitative evaluation of GMs.

As far as we know, it is the first study about GMs in children with Down syndrome.

## 2. Methods

The assessment of General Movements (GMs) is a simple, un-invasive and reliable method for neurological evaluation of preterm and term infants. GMs can be observed in fetuses as young as 9 weeks postmenstrual age and after birth until about 4–5 months.

GMs are gross movements, involving the whole body, lasting from a few seconds to several minutes or longer. They wax and wane in intensity, force and speed, and have a gradual beginning and end. Arm, leg, neck and trunk are involved in variable and complex sequences of movements producing the impression of fluency and elegance [10].

During the first 2-month post-term, GMs are commonly referred to as Writhing Movements (WMs). They are characterised by small to moderate amplitude and by slow to moderate speed, and are elliptical in form; this component creates the impression of a writhing quality of movement [11].

At the age of 6 to 9 weeks post-term, the form and character of GMs of typically developing infants changes from a Writhing type to a Fidgety pattern. Fidgety Movements (FMs) are circular movements of small amplitude and moderate speed, they are continual in the awake infant, except during focused attention [12]. They may be concurrent with

other gross movements, such as kicking, wiggling–oscillating and swiping of the arms or ‘pleasure bursts’.

Fidgety movements are transient in healthy infants: initially, they occur as isolated events at about 6 weeks corrected age, gradually increase in frequency, coming to full expression between 9 and 13 weeks corrected age, and then decrease once again between the ages of 14 and 20 weeks post-term corrected age, when infants show other patterns of motor activity, such as manipulation, voluntary movements, reaching, touching, grasping, axial rolling, and antigravity movements [11–14].

The results of GMs evaluation highly correlate with the neurological outcome: a normal quality of GMs consistently predicts a normal neurological outcome, while an abnormal quality of GMs predicts an abnormal outcome.

The method’s sensitivity is 94%, and similar in all ages. The specificity is age-dependent: it is low during the first 2 months and reaches 82% to 100% at 3-month post-term. There is a robust character of this method: reported inter-observer agreement is between 78% and 98%, and reported kappa coefficients are between 0.84 and 0.92 [15,16].

The effects of many neurological and not-neurological diseases on spontaneous motility’s character have been analysed in previous studies [17–22].

We performed a prospective study. During the years 1999–2003, 53 children (25 males and 28 females), 23 infants with DS (Down group), 30 healthy full-term infants (control group) were selected.

Down group (13 males and 10 females): all infants had gestational age ranged 36–41 weeks postmenstrual age (mean 38.2 weeks; S.D. 2.76); birth weight between 1759 and 3050 g (mean 2667.3 g; S.D. 413.13); 13 children had normal delivery, 10 had Caesarian delivery; APGAR score ranged 7–9 at 1 and 5 min. All infants from the Down group had confirmed diagnosis of full trisomy 21 by cytogenetic findings.

Control group (12 males and 18 females): all infants had gestational age ranged 38–40 weeks postmenstrual age (mean 38.9 weeks; S.D. 0.87); birth weight between 2800 and 3400 g (mean 3081 g; S.D. 202.39); normal delivery; APGAR score ranged from 8 to 10 at 1 and 5 min.

Informed consent was obtained for all children who participated to the study. Statistical analysis was performed using the Students’ *t*-test.

### 2.1. Study design

For each child, from 1st to 6th month corrected age, we performed an average of two brain ultrasound scans (range 1–3); standardised repeated neurological examination (according to the Dubowitz and Dubowitz [23] protocol during the preterm period and according to the Prechtl [24] protocol at term age) and behavioural examinations (according to the Brazelton [25] protocol). Later development was checked by means of the Amiel-Tison and Grenier [26] examination or by use of the Touwen [27] criteria.

### 2.2. Observation and evaluation of spontaneous movements

According to Prechtl [13,14,28], 1-month-old infants were evaluated before discharge from the hospital performing 1-h videotape recording of the spontaneous motor activity.

After discharge, 10–30-min videotape recordings were acquired every 4 weeks in the outpatient clinic. All the recordings were made about an hour before feeding and during active wakefulness (States 3–4 according to Prechtl and Beintema [29]). Infants were undressed or partially dressed (i.e., with a body vest and/or nappy), lying supine and unstimulated. The video camera was positioned approximately 1 m above the cot at an angle of 45°.

During the playback of the video recordings, 3 to 4 GMs each lasting 30 to 120 s, excluding crying, fussing, or suckling periods, were selected as typical for each particular age. A collection of GMs at different ages was then assembled and stored on a separate tape for each infant [28,30].

GMs were analyzed in accordance with the previous studies [21,28,31,32] in two ways: a qualitative assessment of normality or abnormality of GMs, based on a global visual gestalt perception of the complexity, fluency and elegance or monotony and awkwardness of spontaneous movements; and a semi-quantitative estimation of GMs, assessing separate aspects of the spontaneous motor activity: amplitude, speed, sequencing, fluency and elegance, onset and offset.

For the semi-quantitative evaluation of the spontaneous motor activity, we used a modified version (Table 1) of the evaluation sheet used by Ferrari et al. [21].

Table 1

Semi-quantitative estimation of GMs has been assessed evaluating separate aspects of the spontaneous motor activity: amplitude, speed, sequencing, fluency and elegance, onset and offset, movement character

| Evaluation sheet of GMs quality |  |   |
|---------------------------------|--|---|
| Amplitude                       | Predominantly small ranged                   | 1 |
|                                 | Predominantly large ranged                   | 1 |
|                                 | Small and large, no intermediate range       | 1 |
|                                 | <i>Variable</i>                              | 2 |
| Speed                           | Monotonously slow                            | 1 |
|                                 | Monotonously fast                            | 1 |
|                                 | Slow and fast, no intermediate               | 1 |
|                                 | Invariable                                   | 1 |
|                                 | <i>Variable</i>                              | 2 |
| Sequencing                      | Only synchronized movements                  | 1 |
|                                 | Disorganized                                 | 1 |
|                                 | Monotonous sequence within single GM         | 1 |
|                                 | Some body parts are not involved in the mov. | 1 |
|                                 | Repetition of same sequence from GM to GM    | 1 |
|                                 | <i>Variable</i>                              | 2 |
| Fluency and elegance            | Not fluent, no rotations                     | 1 |
|                                 | Not fluent, few rotations                    | 1 |
|                                 | <i>Fluent and elegant, many rotations</i>    | 2 |
| Onset and offset                | Abrupt                                       | 1 |
|                                 | Minimal fluctuation in intensity             | 1 |
|                                 | <i>Smooth crescendo and decrescendo</i>      | 2 |
| Total                           |  |   |

For each item, we scored 2 points for an optimal performance (indicated in tables with the Italic font stile), and 1 point for a non-optimal one (indicated in tables with the regular font stile). Total score (ranged 5–10) was obtained summing up each item's score.

According to the optimality concept [33] on each item (Amplitude, Speed, Sequence of movements, Fluency and smoothness, Onset and offset), we scored 2 points for an optimal performance (indicated in tables with the *Italic font style*), and 1 point for a non-optimal one (indicated in tables with the *regular font style*).

Total score (ranged 5–10) was obtained summing up each item's score.

Qualitative and semi-quantitative assessments of GMs were achieved blindly for all recordings by each of the co-authors. All the observers have a long clinical experience in neurological assessment of preterm and term newborns, and have a 5-year experience with the evaluation of spontaneous motor activity (GMs). The assessors were tested to ensure they had reached acceptable level of inter-rater reliability for scoring of GMs (weighted  $k > 0.80$ ). Criterion test scores of individual assessors ranged from a kappa value of 0.87 to 0.98 indicating excellent agreement. Gestalt perception was recalibrated each week by reviewing “gold standard” tapes of normal and abnormal GMs [28,30].

Previous studies [14,19] demonstrates that the test–retest reliability after an interval of 2 years is 1.0 for the global judgment (normal vs. abnormal) and 0.85 for the analysis of details (amplitude, movement character, sequencing, fluency and elegance, etc.).

### 3. Results

Normal brain ultrasound scan was found in all children with no known motor problems. Fifteen infants with Down syndrome had normal brain ultrasound scan at birth and eight had transient periventricular hyperechogenicity.

The neurological evaluations in the control group did not show any abnormality; all the children with Down syndrome showed slight to moderate generalised hypotonia.

#### 3.1. GMs in children with Down syndrome

##### 3.1.1. From birth to 2nd month corrected age

Qualitative evaluation: GMs were generally characterised by low speed, large amplitude, absence or partial absence of fluency and smoothness; the sequences of arm, leg, neck and trunk movements were generally few variable and complex. No or very few typical elliptical forms of the limb movements were visible. No fast and large extensor movements of the arms or legs were observed. Spontaneous motor activity was scattered during the recording; beginning and end were generally abrupt lacking wax and wane in intensity, force and speed.

Semi-quantitative evaluation: at 1st-month observation, 15% of children with Down syndrome showed an optimal performance at the parameter “Amplitude” while 85% had a non-optimal performance; 16% achieved an optimal performance at the parameter “Speed”, while 84% had a non-optimal one; at the parameters “Onset and offset” and “Fluency and smoothness” 25% of children with Down children had an optimal performance, while 75% had a non-optimal one; 60% showed an optimal performance at the parameter “Sequence of movements”, 40% had a non-optimal performance. Mean total score was 6.69 (S.D. 0.97).



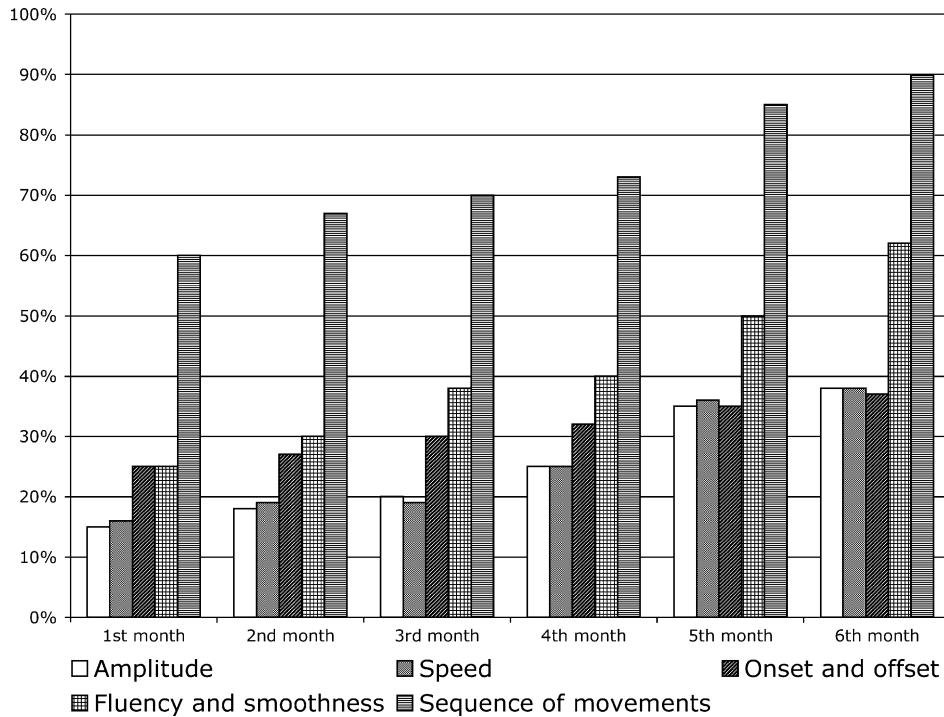


Fig. 1. Children with Down syndrome: for each aspect of GMs (Amplitude, Speed, Onset and offset, Fluency and smoothness, Sequence of movements) percentage of children achieving an optimal performance at each time point is indicated.

showed a decrease in the occurrence of GMs and a clear development of voluntary movements.

### 3.2. GMs in control group children

#### 3.2.1. From birth to 2nd month corrected age

A mean total score of 9.26 (S.D. 1.36) was achieved at first month; five children showed spontaneous motor activity characterised by slight abnormalities (small or large ranged movements, monotonously slow or fast speed, absence of fluency or smoothness), that we considered developmental age-related changes of normal GMs. The sequence of arm, leg, neck and trunk movements were variable and complex in all children.

The 2nd-month evaluation revealed an increase of the mean total scores to 9.73 (S.D. 1.17), GMs giving now the impression of a Writhing quality in almost all infants.

#### 3.2.2. From 2nd to 6th month corrected age

All children showed continual circular movements of small amplitude and moderate speed, concurrent with other gross movements (kicking, wiggling–oscillating and pleasure bursts).

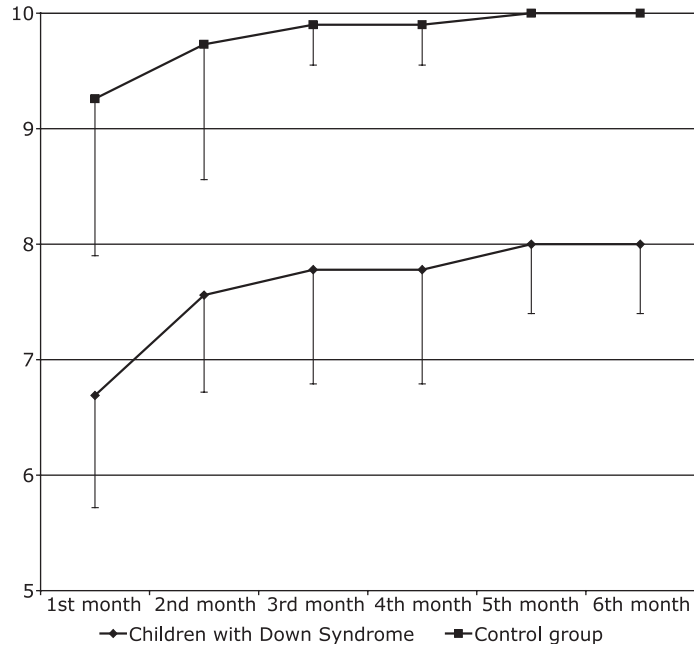


Fig. 2. Mean total scores and standard deviations of the motor optimality score obtained by children with Down syndrome are compared to mean total scores and standard deviations of children with no known motor problems.  $p < 0.0001$  at each time point.

At 3rd and 4th months, mean total score was 9.9 (S.D. 0.35); increasing to 10 (S.D. 0) at 5th and 6th months.

The other patterns of motor activity, such as manipulation, voluntary movements, reaching, touching, grasping, axial rolling, and antigravity movements gradually developed at 15–20 weeks post-term age.

Data from the semi-quantitative evaluation were graphically displayed to obtain growth curves of motor optimality scores (Fig. 2). For each time point, mean total scores and standard deviations of children with Down syndrome were compared to mean total scores and standard deviations of children with no known motor problems (Table 1).

Student's  $t$ -test demonstrated statistically significant differences between the two groups at each observation ( $p < 0.0001$ ).

#### 4. Discussion

The aim of our study was to describe the character of the spontaneous motor activity in children with DS, compared with healthy full-term infants.

Five different patterns of abnormal GMs have been described by Prechtl et al. During the prenatal period and during preterm, term and the first 2 months post-term age: *Poor repertoire General Movements* (PR), characterised by lack of complexity and variability of sequence of the successive movement components; *Cramped-synchronised General*



*Movements* (CS), characterised by rigid and abrupt movements, and lack of the normal smoothness and fluency; *Chaotic General Movements* (Ch), characterised by large amplitude of the movements, without any fluency nor smoothness.

Fidgety movements are referred as abnormal if they are *Absent* (F<sup>-</sup>): FMs are never observed from ages 6 to 20 weeks post-term, or *Abnormal* (F<sub>A</sub>): when their amplitude, speed and jerkiness are moderately or greatly exaggerated [31,32].

Prechtl et al. demonstrated that observation of spontaneous motor activity (GMs) in young infants allows valid predictions about later neurological outcome [14]. The qualitative assessment of GMs during first 2 months (WMs) has an high sensitivity, but its specificity is considerably lower [28]. Many of the earlier GMs abnormalities are transient phenomena and normalise before or at the appearance of Fidgety movements. Abnormal or Absent Fidgety movements are indicative of a poor outcome; Normal Fidgety movements are an excellent marker for a normal neurological outcome [14–16,34].

During first 2 months, GMs of children with Down syndrome are generally characterised by low speed, large amplitude, absence of fluency and smoothness, few variability of limbs movements, absence of small ranged limbs movements and of other gross movements.

During the next months, speed of movement change from low to low-moderate, amplitude change from large to large-moderate, and it is possible to observe small-ranged limbs movements and changes in direction of the movements (as well as other gross movements like kicking, wiggling–oscillating and swiping of the arms) partially creating the impression of fluency, smoothness and complexity.

The typical change in form of GMs recurring at the end of the second and beginning of the third month in children with no known motor problems from Writhing to Fidgety character, has not been observed in children with DS. From birth to 6th-month corrected age, GMs of children with DS become progressively faster, smaller ranged, more fluid and elegant, and sequences of limbs movements more variable and complex, corresponding to a continual and progressive improvement of GMs quality and of the motor optimality score, but it was not possible to observe a clear changing in form (Writhing to Fidgety) of the spontaneous motor activity neither at the end of the 2nd month nor later.

All children with DS show an improvement of their optimality scores during the observation, but it was possible to observe widely dispersed values of motor optimality scores—as indicated by S.D.—at each time point, reflecting the heterogeneity of these children: mean total optimality score at 1st month was  $6.69 \pm 0.97$ ; at 2nd month  $7.56 \pm 0.84$ ; at 3rd and 4th months was  $7.78 \pm 0.99$ ; at 5th and 6th months was  $8 \pm 0.6$ .

The pattern and evolutionary course of GMs in children with DS that we described, as well as the delay in disappearing of the spontaneous motor activity and develop of voluntary movements, can be related to the central nervous system and the peripheral abnormalities that characterize this syndrome. It cannot be considered an abnormal pattern (PR, CS, Ch, F<sup>-</sup>, F<sub>A</sub>) as observed in children with severe encephalopathy and evolution in cerebral palsy, but has to be considered typical of children with DS.

Palisano et al. administered the GMFMD and rated on degree of motor impairment (Mild, Moderate and Severe motor impairment) 133 children with DS to create motor growth curves that describe motor function of children with DS and to compare the rate of

improvement and upper limit of function between children with mild motor impairment and children with severe or moderate motor impairment.

They demonstrated that on average: (1) Motor impairment has a discernable effect on rate of improvement but only a slight effect on ultimate achievement of gross motor function during early childhood. (2) Children with DS do not achieve all the gross motor functions included on the GMFM by the age of 6 years. (3) Scores improve the fastest at younger ages and then level off as the predicted upper limit of gross motor function is approached [8,9].

The results of studies that have examined motor interventions in children with DS are still inconclusive both for short-term and long-term benefits [4,35–47].

According to Palisano et al., children whose motor function is age appropriate or advanced relative to expectations for children with DS may successfully achieve motor goals through play and structured developmental activities, and intervention may appropriately be directed to other areas of development like occupational therapy. Conversely, children whose gross motor function is delayed relative to expectations for children with DS may be less likely to achieve goals without physical therapy interventions [8].

We believe that it is really important to have assessment instruments that can help professionals to make early management decisions, set goals for intervention and evaluate intervention outcome.

Our hypothesis is that GMs quali–quantitative evaluation could be an early marker of motor impairment, and could have implications for decision about if and when motor interventions are needed.

The growth curves of the motor optimality scores show the average pattern of development of the children with DS (compared to children with no known motor problem) based on cross-sectional data. Children with Down syndrome whose growth curve of motor optimality score is delayed relative to expectation could have a later delay in the rate of improvement of gross motor functions. These information could lead professionals and parents to start an earlier and more intensive physiotherapy intervention, and to set outcomes earlier and more effectively than using other standardized criterion-referenced measures. Further studies are needed to confirm this hypothesis.

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