Letters to the editor

‘Developmental impairment that is not immaturity’

SIR–Beckung et al. should be commended for their thorough study. Their results provide a comprehensive picture of gross motor development in Angelman syndrome that takes into account the heterogeneity encountered in this condition. They also confirm the importance of clinical diagnosis. Their questioning of whether the severity of neurological abnormalities seen in Angelman syndrome justifies its inclusion within the framework of cerebral palsy echoes a recent analytic study of this construed concept. Nevertheless, their conclusion that ‘motor problems in individuals with Angelman syndrome mainly represent immaturity uncoordinated movement patterns, similar to what is seen in the early stages of motor development’ needs clarifying. This conclusion implies that they organize their motor behaviour in the same way as individuals without disability who have no experience of the considered motor tasks. It appears to be contingent to the authors’ quantification of motor competence as ‘motor age’, which fails to evaluate a dimension that links motor impairment to function, namely motor control. We studied the kinematics of intersegmental coordination in locomotion in toddlers without disability from their first steps through walking development and in children with Angelman syndrome. In normal ‘mature’ walking, this coordination is characterized by highly stereotyped coupling of elevation angles of the thigh, shank, and foot during the gait cycle, which has been termed the planar covariation. The planar covariation reflects the dynamic integration of postural stability with respect to gravity and forward progression; it correlates reliably with mechanical energy expenditure. For their very first steps, normal toddlers did not show planar covariation of the elevation angles of the thigh, shank, and foot during the gait cycle (Fig. 1Aii). In contrast, in children with Angelman syndrome the covariation of the elevation angles of the lower limb segments was constrained on a plane (Fig. 1Bii) whose spatial orientation significantly differed from normal controls. In addition, they did not show physiological in-phase forward rotation of the shank and foot at the end of the swing phase or in-phase.

Figure 1: (A) Very first steps of a child without disability (age 11 mo). (B) Gait of a child with Angelman syndrome (age 9 y). (Ai) and (Bi) are sagittal kinograms: superimposed segments representing a model of body defined by opto-electronic recording of markers placed on anatomic landmarks (nose, ear, acromion, antero-superior iliac spine, trochanter, knee, malleolus, 5th metatarsal). Two consecutive segments = 20 ms. (Aii, Aiii), (Bii, Biii) show covariation of elevation angles at thigh, shank, and foot during two successive steps at onset of unsupported walking. Sampling rate = 100 Hz. Mean value of each angular coordinate has been subtracted. Data are represented with respect to cubic frame of angular coordinates and best fitting plane (grids) in two different perspectives. Gait cycle paths progress in time in counter-clockwise direction, ground contact and toe-off phases corresponding roughly to the top and bottom of loops, respectively.
backward rotation of these segments after the heel strike while the thigh elevation angle remained constant. We also studied the squatting movement in a group of 10 children aged 7 to 13 with Angelman syndrome and in four children without disability but with minimal experience of unsupported standing (3 to 9 weeks following the onset of walking). The latter performed the movement using motor strategies that were different from that seen in Angelman syndrome. Toddlers without disability either kept their trunk erect and performed a low magnitude knee flexion (Fig. 2a) or they showed marked trunk and lower limb joint flexion, in contrast to the pattern shown by children with Angelman syndrome (Fig. 2c). Both patterns (Fig. 2a,b) could be observed in one child, but no intermediate pattern was seen. The trunk-erect pattern was successfully recorded four times, the global flexor pattern nine times. Moreover, greater intra-individual kinematic variability was found in both tasks in toddlers without disability than in children with Angelman syndrome, suggesting adaptability based on diversity, competition, and choice in the former and predominance of a stereotyped ‘fail-safe’ mechanisms in the latter.

Furthermore, angular orientation of the head was more stable in toddlers than in those with Angelman syndrome, suggesting a mode of control relying on a head-centred referential – as further refined in older normal children but not in Angelman syndrome. Finally, in a study of upper limb and lower limb isometric postural tasks, we consistently found hypersynchronous rhythmic bursts of electromyographic activity in 14 children with Angelman syndrome that was not observed in infants, toddlers, or children without disability. The question of whether patients with developmental impairment organize their motor behaviour in an immature fashion has long been a matter of debate with regard to pathophysiology, diagnosis, and management. These paradigms demonstrate differences between motor patterns observed in Angelman syndrome and ‘immature’ children without disability, that could not be identified with gross motor function measurement.

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References

Figure 2: Sagittal kinograms of children squatting from standing position. (a) Toddler without disability demonstrating trunk-erect pattern. (b) Toddler without disability demonstrating global flexor pattern. (c) Child with Angelman syndrome (age 10y). Sampling rate = 12.5 Hz.
‘Beckung and Kyllerman reply’

SIR–We are grateful for the valuable comments made by Dan et al. regarding our study on motor function in children with Angelman syndrome. We highly appreciate the additional information provided by Bernard Dan and Guy Cheron on advanced kinematic analysis comparing motor control in children developing normally with children with Angelman syndrome.

In the clinical setting of our study we found that distal lower limb spasticity, ataxic-like gait, stiff lower limbs, and the presence of coactivation during locomotion were significantly more frequent in children with Angelman syndrome than in the comparison group of children with retardation of motor function from other causes.

These results indicate that the physiology of pathological locomotion in children with Angelman syndrome is somewhat similar to that described in children with cerebral palsy (CP), and other upper motor-neuron syndromes. However, the dominant impression is of immaturity in execution, on a par with the generally low developmental level.

As collaborators of the Surveillance of Cerebral Palsy in Europe (SCPE) network we apply the definition of CP as ‘a group of disorders, involving a disorder of movement and posture and of motor function, permanent but not unchanging, due to a non-progressive interference, lesion, and/or abnormality in the developing/immature brain’. In our opinion, motor problems in children with Angelman syndrome mainly represent immature discoordinated movement patterns, similar to those seen in the early stages of motor development. All children in this study functioned at a very immature gross and fine motor developmental level. We did also imply in our paper that the specific movement patterns were not purely immaturity. Signs of ataxia and spasticity were more prevalent in the Angelman syndrome group than the comparison group, which may indicate that there are additional dysfunctions specific to children with Angelman syndrome. We also found that the movement pattern was ataxic-like but differed from the dysmetria, dyssynergia, and dysequilibrium seen in individuals with cerebellar ataxia syndromes. Neurological abnormalities were mild, and, in our opinion, did not qualify for a diagnosis of CP.

Difficulties experienced by children in the test probably resulted from central discoordination resulting in difficulties in positioning the body and interacting with the environment. Children seemed to have problems in sensory-motor integration, i.e. deficient interaction between multiple spinal and supraspinal systems. We considered that neurological abnormalities observed in children in our study with Angelman syndrome impeded motor function less than or as much as immaturity did. On the grounds mentioned above, the motor difficulties were insufficient for a diagnosis of spastic diplegic or ataxic CP, although mild neurological signs were present.

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References

Announcement and Call for Papers

International Symposium on Epileptic Syndromes in Infancy and Early Childhood – Evidence-based Taxonomy and its Implications in the ILAE Classification

Date: April 29 – May 1, 2005
Venue: Tokyo Women’s Medical University
Japan
Host: Infantile Seizure Society, Japan
President: Yukio Fukuyama, MD

Early registration: February 28, 2005

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