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ANNOTATION

CHAOS IN THE CLASSIFICATION OF SMA: A POSSIBLE RESOLUTION

Earlier discussions on a comprehensive classification of the spinal muscular atrophies of childhood, mainly to provide a rational basis for gene linkage studies, led to the International Consortium on Spinal Muscular Atrophy producing a compromise between the numerical approach, subdividing the SMAs into types 1, 2 and 3, based on age of onset and age at death, and the classification into three groups based on severity (severe, intermediate, and mild), reflecting the three subdivisions of clinical severity based on the ability to achieve independent sitting and independent standing and walking [1–3].

This classification did not take full account of the variability in severity within each of the three groups and the continuum of clinical severity from the most severe end of the severe group (type 1) to the mild end of the mild group (type 3) [4].

As one might have anticipated, the numerical classification introduced a concept of rigidity which did not reflect the reality of the clinical condition. Cases are being documented of type 1 SMA, with prolonged survival, and some observers are now going as far as to say that one cannot predict survival at all in type 1 SMA as one cannot tell whether the individual infant may survive 1 yr or 30! This is patently nonsense and anyone looking at the infant with classical severe spinal muscular atrophy ("Werdnig–Hoffmann disease") and taking note of his compromised respiratory function, which is the only life threatening component of the disease, can rapidly assess the likelihood of survival beyond infancy or early childhood.

This posed the question whether the long-term survivors of type 1 SMA (who had never achieved the ability to sit unsupported) were either milder cases within this group, with correspondingly better respiratory function and better prognosis, or alternately represented some entirely different clinical phenotype, possibly with a different genetic basis. This was of obvious importance to resolve in view of current attempts at genetic counselling and prenatal diagnosis in relation to 5q11-13 classical SMA.

In view of a number of these long surviving cases identified by Rudnik-Schöneborn and Zerres (personal communication) as part of the very large genetic study undertaken by Zerres in Germany [5], I visited Cologne in order to personally assess some of these type 1 SMA cases with long survival to try and resolve this diagnostic quandary. The answer turned out to be the first suggestion of milder cases within the type 1 classification. Three children were assessed in detail (scattered over a distance of 600 km in West Germany, which we covered in a weekend). One was able to maintain a sitting posture in a wheelchair, with appropriate supportive propping, but had considerable respiratory problems and had had several life-threatening attacks of pneumonia. Although she had survived until 12 years of age, her prognosis was obviously poor, in view of the threat of further pneumonic episodes and a very poor respiratory reserve. She was in fact identical in appearance to some of the cases I have documented that I have called borderline in severity between the severe and the intermediate group [6, 7].

The other two patients also straddled the dividing line between the type 1 (severe) and the type 2 (intermediate) group. One, now aged 10 yr, had in fact achieved independent sitting, but only intermittently for short periods, on looking through family photographs. The other child had not achieved independent sitting but had been able to kneel unsupported at an early age (family photograph), and obviously had good function in the spinal muscles and was now, at the age of 9 yr, able to maintain a sitting posture independently for short periods.

I reflected on how one could possibly intro-

duce into the somewhat rigid mathematical concept of the three numerical types of SMA, some flexibility to accommodate the variable clinical pattern. Somewhat tongue in cheek, I suggested that if people preferred numerical types, rather than clinical descriptions, and wanted to treat clinical disorders as rigid mathematical formulae, they should perhaps consider carrying this approach further and subdividing the cases on a decimalized system, in order to reflect the difference between the infant with type 1 SMA, with severe paralysis at birth and early respiratory and bulbar difficulties and an overtly poor prognosis for survival (type 1.1) and the infant at the other end of the type 1 scale, with onset of weakness, perhaps at 3–5 months of age, almost achieving the ability to sit unaided, but not quite, and having a more reasonable respiratory reserve, although still somewhat compromised (type 1.9). There was an obvious distinction between these two extremes of type 1 cases. One could adopt the same approach to reflect the variability within the intermediate group (type 2), recognizing the infant at the severe end of the group, able to sit unsupported, but only just, and being very floppy and insecure (type 2.1), in contrast to the infant at the other end of the scale, able to sit very well without support, with a straight spine, and able to maintain this sitting posture for extended periods (type 2.9) and in addition also able to take some weight on his legs when supported, but not to maintain independently a standing position. One can also recognize the same variability within the mild group of SMA (type 3). At the severe end is the child who is just able to stand unsupported and take a few steps unaided, but is very unstable (type 3.1), whereas at the other end is the child who is walking well, able to go up and down steps, able to get up from the floor with minimal Gowers' sign and only showing a very minimal disability (type 3.9) (see Table 1). (Type 4.0 would represent normal function.)

Table 1. Classification of Childhood SMA

Type 1	Severe (variable)	Type 1.1–1.9
Type 2	Intermediate (variable)	Type 2.1–2.9
Type 3	Mild (variable)	Type 3.1–3.9
	(continuum from severe to mild)	
("Type 4.0" = normal)		

Within each type one could then quite readily define one's own subdivisions, perhaps initially finding an approximate midpoint within each type and then trying to reflect the variability on either side of this, in a practical way. Thus, within the severe group one might categorize type 1.5 as the child with classical features of severe SMA, inability to raise the legs against gravity or maintain the head posture either in the supine or prone position, but having no difficulty with feeding and swallowing, no accumulation of pharyngeal secretions and no obvious distress. In view of the presence of intercostal weakness and the costal recession and mainly abdominal breathing, this child is of course still at risk of pneumonia or respiratory compromise following mild upper respiratory tract infections. Similarly one can categorize the type 2.5 within the intermediate group as the child who is sitting well without support, has a straight back, but is unable to maintain this for any length of time and is also unable to take any weight on the legs.

One still needs to take account of the fact that within each group the ultimate prognosis for survival depends primarily on the respiratory function and not necessarily on the motor ability. Thus, within the type 2 group in particular, there are some cases that will be surviving into adolescent and adult life, whereas others may die of pneumonia in early childhood or in their teens and one could again try and identify these particular cases as a result of assessment of their respiratory function and reserve and also their liability to recurrent bouts of chest infection. However, it is extremely difficult if not impossible to predict prognosis for survival, owing to the unpredictability of potentially life-threatening bouts of pneumonia.

One also needs to take note of the fact that a child who is freely ambulant (say type 3.5), and apparently stable, may yet lose the ability to walk unaided during the prepubertal growth spurt, owing to changes in body dimensions in relation to a static muscle power, and may need to be remobilized in calipers. Conversely the child who is unable to stand or walk independently, but has fairly good motor function and is sitting well (say type 2.7), may achieve the ability to walk with calipers and even to do so over long distances and may subsequently even improve in strength.

I hope that this approach to the classification of the childhood SMAs, which takes account of the extreme variability within the condition, will help resolve the conflict between the various enthusiasts with a pathological attachment to their own individual classifications

(including, of course, myself). It is not my intention to introduce an even greater degree of mathematical precision into the numerical classification; the individual decimal points are only meant to reflect the continuum and variability within each group. Perhaps one only needs to visualize the two extremes (.1 and .9) and a midpoint (.5) and the fact that there is something in between. Under no circumstances whatsoever should one contemplate a second decimal point.

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