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Editorial

Spinal Muscular Atrophy Revisited

The mills of God grind slowly ..... but they are no patch on the medical profession.

In 1902 Beevor [1] drew attention to the bell-shaped chest with paralysis of the intercostal muscles and sparing of the diaphragm in an infant with severe limb weakness who subsequently died at six months of age. Autopsy showed loss of anterior horn cells of the spinal cord. Three previous infants had been similarly affected and died by six months of age.

The first clinical picture I have found of the severe form of SMA showing the bell-shaped chest with wasting of the intercostals and the prominent abdomen and paradoxical movement of the diaphragm was in a personal copy of the second edition of the paediatric textbook of Jonathan Hutchinson published in 1910 [2] was subsequently able to verify the inclusion of the same figure in the first edition of the book in 1904 (Figure 1).

Figure 1

Following the location of the gene for SMA in 1990, a meeting of clinicians and geneticists was convened in New York to promote further collaborative studies in identifying the gene [3].

The committee decided to classify the different severities of SMA on the basis of the age at onset of symptoms and the age at death. I strongly disagreed with this approach as I thought it more important to relate the severity to the degree of muscle weakness, and also drew attention to the cardinal feature, unique to SMA, which is the paralysis of the intercostal muscles with sparing of the diaphragm [4]. I was unable to convince them that this was possible on simple clinical observation without special investigation such as X-Ray screening, which was likely to give a spurious result. Figure 2 shows three infants with severe SMA with the

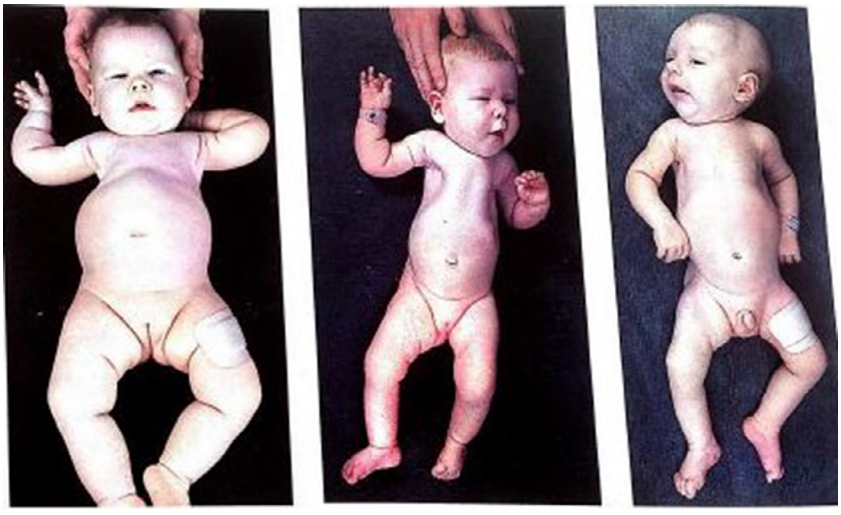

Figure 2

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bell-shaped chest with inactivity and wasting of the intercostals and sparing the diaphragm [4].

The committee were intent on dividing the cases into three numerical "types", based on the achievement of sitting unsupported (type 2) and standing and walking (type 3). The severe cases with inability to sit unsupported (type 1) were subsequently subdivided into type 1A or 1B on the basis of the age of onset. From a clinical point of view it matters not whether an infant is subclassified into type 1A or 1B on the basis of the age of onset, as the only prognostic factor for survival is the degree of respiratory compromise [5,6].

In the current issue of the Journal, Ropars and her colleagues have revisited the intercostal weakness in severe SMA and suggested measuring chest circumference at the nipple line in comparison with the relatively stable or slowly increasing head circumference. The basic question is whether all this accumulated data on a small and variable sample will give you any more information than simple observation of the striking change in the appearance of the chest after effective treatment, or simply plotting the change in chest circumference after treatment.

Victor Dubowitz
Editor-in-Chief

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