Conclusions

Although treatment for asthma has improved considerably over the past 30 years, many sufferers and carers continue to experience problems in their everyday lives. This may in part reflect the difficulty of fully controlling the disease; further work is certainly needed to examine the precise relationship between symptom control and social and emotional problems.23

In the meantime, parents state that the impact of asthma on themselves and their children is seldom fully recognised.7 They often encounter scepticism or incredulity when they describe their problems; indeed, they are often blamed for causing those problems themselves. A better appreciation of the social and emotional impact of asthma is essential if children and their families are to receive the care and assistance they need.

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7 Nocon the Booth T. The social impact of asthma. University of Sheffield: Joint Unit for Social Research, 1989.

Hyperexplexia or stiff baby syndrome

The stiff baby syndrome or hyperexplexia is an unusual neurological disease, occurring via dominant autosomal transmission, and distinguished by a permanent hypertonia that is heightened by the slightest stimulus. The diagnosis can further be ascertained by the clinical and electromyographic aspects, as well as the occurrence of a family history.

The evolution of the disease is benign with myorelaxing treatment. The recognition of this syndrome is of great importance, in order to avoid an erroneous diagnosis of epilepsy, to warn the parents against the risk of recurrence in case of later pregnancies, and to indicate the necessity of constant supervision during the neonatal period as attacks of hypertonia may lead to apnoeas that can endanger the child’s life.

Hyperexplexia was described for the first time in 1962 by Kok and Bruyn in 29 members of one family.1 The disease was characterised by the occurrence of a hypertonia from birth, which became less pronounced during the first year of life but could later lead to repeated falls. Those characteristics were later reported by Suhren et al on 25 members of a family.2 In 1972, Klein described a family of 10 who presented with a syndrome distinguished by attacks of hypertonia heightened by the slightest physical stimulus and in whom an electromyogram showed a persistent activity even on resting; this activity vanished on treatment with diazepam.3 In all cases a muscular stiffness appearing from birth is reported, which gives the child a peculiar attitude described as ‘fetal’, and there is a flexion of the forearms and legs and closed fists. The expression of the child is also characteristic: the stare is fixed and gives the child an expression of anxiety. The hypertonia diminishes during sleep but increases with the slightest psychic or tactile stimulus. Attacks of hypertonia can involve respiratory muscles and lead to apnoeas, endangering the child’s life. Digestive troubles, such as vomiting, are also reported. Vomiting is, in most cases, in association with a hiatial hernia. It is also noted that umbilical, inguinal, and diaphragmatic herniae are more frequent; they are attributed to the hypertonia.

Electroencephalography is normal but electromyography gives a characteristic result and shows an almost permanent muscular activity with, however, periods of electric quietness. The nerve conduction velocity is normal. The evolution of the disease is characterised by a delay in reaching motor ‘milestones’ (for example, walking). The hypertonia is reduced during the first two years, but abnormal reactions such as involuntary starts remain after the slightest stimulus. There is neither mental nor neurological deficit.

It is important to distinguish the stiff baby syndrome from other neurological diseases manifest by an increase in muscle tone. First, this syndrome must not be confused with the stiff man syndrome described by Gordon et al in 1967.4 The stiff man syndrome is not hereditary and appears at the age of 40 to 60; evolution is slow and progressive. It is distinguished by a permanent contracture, an intense dysphagia, the electromyogram shows a permanent electric activity even during rest, and it is magnified by the slightest tactile or nociceptive stimulus.5 The Isaacs-Mertens syndrome, usually occurring around the age of 30, and for which neonatal forms have been reported, presents a mostly distal hypertonia, accompanied by fasciculations; the
electromyogram shows a permanent activity on resting as well as numerous discharges. The syndrome described by Stevens in 1965 under the name 'Jumping Frenchman of Maine' is different, as violent starts, produced by the slightest stimulus, are associated with an echolalia and an echopraxia. Gilles de la Tourette's syndrome chiefly presents spasmodic twitchings and startling reactions. Several other conditions can produce abnormalities in tonicity: among them are the encephalomyelites, Creutzfeldt-Jakob disease, metoclopramide or strychnine poisoning, and myoclonic epilepsy. The syndrome shows a permanent activity on resting as well as numerous discharges. The syndrome described by Stevens in 1965 under the name 'Jumping Frenchman of Maine' is different, as violent starts, produced by the slightest stimulus, are associated with an echolalia and an echopraxia. Gilles de la Tourette's syndrome chiefly presents spasmodic twitchings and startling reactions. Several other conditions can produce abnormalities in tonicity: among them are the encephalomyelites, Creutzfeldt-Jakob disease, metoclopramide or strychnine poisoning, and myoclonic epilepsy.

The physiopathological mechanism of hyperexplexia is controversial: Markand thinks that the hypertonia is linked to a hyperactivity of cortical neurons, but Suhren et al and Morley et al invoke an abnormality in the inhibitory system of the brain stem. Andermann et al think that the problem is with the serotoninergic system. Myorelaxing treatments such as diazepam are efficient as they enable a lowering of the hypertonia. There can be a reappearance of the symptoms when the treatment is stopped to carry out electromyography.

The recognition of the stiff baby syndrome is of great significance in order to avoid an erroneous diagnosis of epilepsy and consequent treatment with anticonvulsants. Recognition is also important so that treatment with myorelaxants can be started to avoid the abnormal startle reactions that produce falls in older children. It is also important to warn the parents about the risk of recurrence of the disease, and to indicate the necessity of constant supervision during the neonatal period, as attacks of hypertonia can lead to serious apnoeas endangering the child's life, although the evolution of the disease is benign when treated.