

Virgin's role in the story is that of a mother who uses medicine to treat her child,<sup>28</sup> her selection of a remedy recommended by the most prominent medical authorities could scarcely have been more appropriate. The cure would still have been considered a miracle, although based on medical principles. Moreover, by using this device, Chaucer would have appealed to the intellectual capacities of his audience, and thus would have attracted new interest into his retelling of a familiar legend.

In any event, an alternative answer to the problem posed by Chaucer's use of the word grain may be found in medieval medical writings. A scholarly consensus regarding the meaning of this object may never be achieved, and it has even been proposed that Chaucer made his references to the grain intentionally vague in order to accomplish diverse literary and aesthetic goals.<sup>29</sup> Whatever his purpose, the ambiguity of this term has resulted in the study of materials hitherto ignored. Therefore, it may be hoped that the grain and other puzzling elements in Chaucer's works, like the perturbations of a known planet, will continue luring scholars to explore the unknown.

<sup>28</sup> Beichner, loc. cit., p. 307.

<sup>29</sup> Steadman, loc. cit., pp. 389-91.

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### THOMSEN AND MYOTONIA CONGENITA

ASMUS Julius Thomas Thomsen (1815-1896) is a unique figure in medical history in that he made the first definitive description of a disease entity, myotonia congenita, from which he suffered and which could be traced through five generations of his family. Thomsen himself believed that the first description of myotonia had been made by Charles Bell in 1830 in his work *The Nervous System of the Human Body*. Neurologists now seriously doubt whether Bell was referring to the same condition, since his account is more applicable to narcolepsy (Bell and Purdon Martin 1947).

Thomsen was born in Brunsholm in Denmark which has since become part of Germany. He studied medicine at Kiel, Copenhagen and Berlin and qualified in 1839 after presenting his thesis on dipsomania. He finally settled in practice in Kappeln where he was also a member of the Board of Health. He was a gifted lyric poet and several of his poems were set to music by Marschner. Some of these contain references to his affliction which is disguised under the terms 'gout' and 'rheumatism' (Hirsch 1934). In Casper's *Quarterly Journal* (1865-66) he wrote a short contribution on abortion and upon the toxic effects of camphor. In an earlier paper in Oppenheim's *Medical Journal* (vol 47) he wrote an article on 'Cinchonium Sulphuricum'. This was a strange coincidence in that quinine was to become one of the specific therapeutic agents in the myotonic disorder which he was later to describe.

He was, however, 61 years old before he published his original description of the 'deep rooted' hereditary muscular disorder which affected him and some of his ancestors (Thomsen 1876). He says of his myotonia:

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In my case it was the first symptom which showed up for as long as I can remember. As a boy I was often unable to get up from a chair immediately when I was called unexpectedly. If I got up quickly my legs were attacked by this tonic cramp which frustrated every effort to move away. If I summoned up all my willpower to force my legs to move I fell down and lay there for some time as stiff as a board unless I could manage to grasp some supporting object. If I had the time to wait until the cramp eased then the will exerted itself very gradually and almost imperceptibly over the network of nerves. When everything was functioning properly again I was just as nimble as other boys of my age.

The muscular cramps embarrassed and humiliated him throughout his life. 'I often had to put up with scoldings and even detention from the people who looked after me and who had no sympathy or understanding of the condition.'

He was convinced that the secretive nature with which his muscular disorder was regarded by his family had a damaging effect upon his whole character development. His intense sensitivity about his muscular disorder is expressed in the following extract.

Everybody wants to be judged fairly since he knows that what he feels to be an illness will not be seen as this by others but all too often is seen as rudeness, lack of breeding, bad manners. Because of this he is the object of disdain or hurtful mockery. So the person concerned tries to keep secret in every possible way what troubles him. . . . When a game of hide and seek like this is played from childhood it is plain to see that a reflection is cast from it on to the whole mental sphere which in the end must produce many shadows. The author of this article himself has been a victim of this weakness and speaks from experience.

It was the inferred accusation that one of his affected sons was malingering his way out of military service that finally provoked Thomsen to publish his article on myotonic cramps. Thomsen had five affected sons, one of whom died in infancy and another at 10 years from diphtheria. Of the three remaining sons, the eldest was mildly affected by myotonia, but the two younger boys were severely afflicted. The youngest, a farmer, applied to be exempt from military service. Several sworn statements were made by Thomsen and other medical colleagues to the effect that the boy had a hitherto undescribed muscular disability which rendered him unfit for military service. In spite of this evidence, the young man was called up for two months' trial period in the army at Rostock. Here the boy came under the care of Dr. Rothe, a consultant at the University Clinic in Rostock. Muscle biopsy was carried out by Professor Ponfick and the presence of this disorder of muscular relaxation was confirmed, after which Thomsen's son was released as unfit for military service. Had it not been for the intense indignation which Thomsen felt about this whole incident it seems doubtful if he would ever have publically disclosed the familial disorder about which he was so sensitive.

In some later correspondence prior to publication of his paper Thomsen referred to the disorder as 'ataxia muscularis' because of the disturbances of balance which the tonic cramps produced. This title was criticized by Professor Bartells who pointed out to Thomsen,

by the word ataxia is meant a disturbance of nervous pathology in the co-ordination of voluntary movement, as for example in *tabes dorsalis*, in the staggering gait in many diseases of the 'small brain' and finally like drunkenness, but not prevention of voluntary movement caused by cramp I would advise you to call it 'Tonic cramps in voluntary muscles as a result of hereditary disposition'.

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This title was finally adopted by Thomsen (1876). Later, Strümpell called the condition myotonia congenita whilst Westphal gave it the eponym of 'Thomsen's Disease'. Both were accepted and given official recognition by Erb in his celebrated monograph on the disorder (Erb, 1886).

#### THOMSEN'S VIEWS ON AETIOLOGY AND PSYCHOSIS

Thomsen believed that the muscular spasms and cramps arose from a failure of the 'will' to assert itself over the muscles and that the ultimate cause lay in the central nervous system.

The root of the illness is, I suppose, certain to be found in the central nervous system in the brain itself, in the part where the will operates whence it is unable to make its way through the nerves of movement to the organs which it wants to set in motion.

This view was largely determined by Thomsen's personal observation that muscular spasms were frequently provoked by sudden changes in his emotional state. 'Fear and anger can aggravate the excitable disposition and even great jubilation can produce it.' He further describes how fear of ridicule or even thinking about the possibility of a muscular spasm whilst in a social setting might provoke a cramp. 'From this it can be assumed that the roots of the illness are not to be found in one single part, but in the whole of the central nervous system.'

Thomsen's views on the neurogenic origin of the disease were further reinforced by the history of mental illness in several of his ancestors and he came to believe there was a direct relationship between the muscular disorder and the mental disorder.

As far as the mental disturbance is concerned which occurs fairly often, there is a great similarity as there is in the muscular complaint. In the nosological system it cannot be classified under silliness. It is more a kind of imbecility, confusion of ideas combined with a tendency to talk drivel with vacant brooding. It has more in common with a certain kind of mental weakness which occurs in old age.

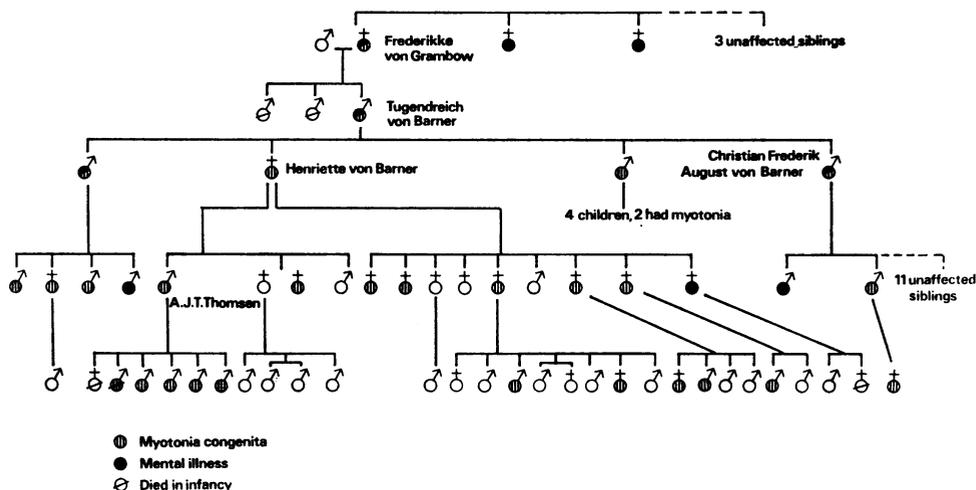
Thomsen's grand-nephew, Nissen, reviewed the family pedigree over seven generations and concluded that the association was fortuitous. '... psychic disturbances can often accompany the picture of Thomsen's disease but do not belong to the complex of symptoms.' (Nissen 1923).

The association of Thomsen's disease and psychosis has recently been reviewed (Johnson 1967) and a further family in which this association was present in some of its members was reported. Two other independent families have also come to the author's attention in which myotonia congenita and hereditary psychosis existed in different members. None of these clinical studies can, however, uphold Thomsen's assertion that the muscular disorder and psychosis are fundamentally related.

#### THE THOMSEN PEDIGREE (see diagram p. 193)

Thomsen traced members of his family afflicted with myotonia congenita back to his maternal great-grandmother, Frederikke von Grambow, who died in 'puerperal mania'. Her only surviving son Tugendreich von Barner had severe myotonia. He became a customs officer but developed a psychosis in middle life and was retired.

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He had four children. The two youngest were severely affected with myotonia; one was 'limited mentally' whilst the other developed a psychosis in middle life. The eldest child had myotonia congenita and also developed a psychosis in middle life. This brought him into conflict with the law, but he was eventually pardoned as a consequence of his mental state. He had three sons, two of whom were affected with myotonia whilst the third was free of it. He suffered, however, from recurrent melancholia and committed suicide at the age of 29.

Thomsen's mother, Henriette Nicoline von Barner, was the remaining child of Tugendreich von Barner. She had slight myotonia and was a highly intelligent woman. She married twice and had 13 children, 7 of whom were affected with myotonia. None developed any mental illness although Thomsen commented upon their tendency to depression.

Nissen (1923), a grand-nephew of Thomsen, investigated a further two generations of the family and in all found 64 affected with myotonia out of a total 315 members. Thomsen always maintained that the disease was milder in his contemporaries than in the earlier generations. This was not borne out by Nissen nor by even later investigators of the family (Thomasen 1948) who found many new cases of myotonia in the descendants of Christian Frederik August von Barner, a brother of Thomsen's mother.

Myotonia congenita is transmitted as a Mendelian dominant in the Thomsen family. It is not sex-linked and has had no effect on fertility. Recessive patterns of transmission were reported in independent families by Thomasen (1948) whilst Becker (1966) in an extensive review of 150 cases found this much more common than the dominant form. According to Becker, myotonia due to the recessive gene does not become manifest until 4-6 years and is more severe than the dominant form.

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### CONCLUSION

Thomsen's description of this rare muscular disorder remains unchallenged in its clinical accuracy and has the stamp of one who has a personal intimacy with the disease. Likewise, the hereditary nature of the disorder has been confirmed, although in recent years the recessive, as well as the dominant modes of transmission have been recognized. Thomsen's views that the origin of the myotonia congenita lay within the central nervous system must, however, now be dismissed in view of the experimental evidence, reviewed by Denny-Brown and Nevin (1941), that the cause of the spasm is some idiosyncrasy of the muscle membrane itself. Indeed, more recent work (Hoffman *et al.* 1966) suggests that the electrical instability of the muscle fibres is due to 'sodium leak' through the membrane. The condition is therefore a genetically determined bio-electrical abnormality of voluntary muscle fibres.

The close association of myotonia congenita and psychosis, which so much concerned Thomsen, has been regarded as fortuitous by subsequent investigators.

The argument has been put forward that myotonia congenita, dystrophia myotonica and paramyotonia are 'one and the same disease' (Maas and Paterson 1939). All the recent studies, however, dismiss this unitary hypothesis and confirm that the disorder described so accurately by Thomsen is a distinct clinical and genetic entity.

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