

Charcot-Marie-Tooth Disease and the Spine

A patient guide

In brief: Charcot-Marie-Tooth disease, commonly abbreviated to CMT, is the commonest inherited disorder of the peripheral nerves, affecting roughly one person in every two and a half thousand. It is a group of genetic conditions in which the long nerves that run out to the hands and feet gradually deteriorate, so that the muscles they supply become weak and wasted and sensation is dulled. Most descriptions of CMT concentrate, understandably, on the high arched feet, the clawed toes and the weakened hands that are its everyday hallmarks. This page takes a different and narrower view. It looks at CMT through the eyes of a spinal surgeon, because the condition carries a notable and well documented association with curvature of the spine. Scoliosis, a sideways and rotational bending of the spine, and kyphoscoliosis, the same deformity combined with an exaggerated forward stoop, occur in CMT considerably more often than in the general childhood population. The reason lies in the very nature of the disease: when the nerves that drive the muscles supporting the spine become unreliable, the delicate muscular balance that keeps a growing spine straight is disturbed, and a deformity can develop. This is what is meant by a neuromuscular curve, and it behaves differently from the common adolescent scoliosis that arises for no known reason. The central idea of this page is that the spine in CMT is shaped by a peripheral nerve problem, and that this colours how the curvature appears, how it is followed over time, and what is weighed when correction is considered in someone whose nerves are already compromised.

What Charcot-Marie-Tooth disease is, and why the spine is involved

Charcot-Marie-Tooth disease is named after the three physicians who described it in the late nineteenth century, and it is also known as hereditary motor and sensory neuropathy. It is not a single illness but a family of inherited neuropathies that share a common theme: the slow, length dependent degeneration of the peripheral nerves, the cabling that carries signals between the spinal cord and the muscles and skin of the limbs. Because the longest nerves are affected first, the feet and lower legs tend to declare themselves before the hands. The result is a characteristic picture of high arched feet, hammered or clawed toes, wasting of the small muscles below the knee, weakness of the ankles, reduced reflexes and a blunting of sensation in the feet and fingers.

The connection to the spine follows naturally once the underlying problem is understood. The spine is held upright and kept straight not by bone alone but by a continuous, finely judged effort from the muscles of the trunk: the long muscles running either side of the backbone, the deeper segmental muscles, and the abdominal wall. These muscles work in opposing pairs, and a straight growing spine depends upon their pull being roughly balanced across the midline. In a condition that interferes with the nerves supplying muscle, that balance can be lost. If the muscles on one side of the trunk pull a little more strongly or a little more steadily than those on the other, a young spine that is still growing can be drawn gradually into a curve. This is the essence of a neuromuscular scoliosis, and it is the mechanism that links a disease of the peripheral nerves in the feet and hands to a deformity of the spine.

It is worth being clear that not everyone with CMT develops a spinal curve, and that the great majority of the disability associated with the condition relates to the limbs rather than the back. The spine is, nonetheless, a recognised site of involvement, and the association is strong enough that curvature of the spine in a child is, in some forms of CMT, among the features that draw attention to the underlying neuropathy in the first place.

How common spinal deformity is in CMT

The association between CMT and curvature of the spine is one of the more consistent findings in the orthopaedic literature on the condition, even if the precise figures vary from study to study. Reported rates of scoliosis in CMT generally fall between roughly a tenth and a little under two fifths of patients, which is appreciably higher than the small percentage seen in the general adolescent population. The variation reflects differences in how the studies were conducted: the ages of the people examined, the particular genetic subtypes included, and the angle at which a curve was counted as a deformity.

The most frequently cited series give a useful sense of scale. A retrospective study of one hundred children with CMT found spinal deformity in well over a third of those with adequate radiographs, made up of scoliosis, kyphoscoliosis and pure kyphosis; among those followed to skeletal maturity, the proportion with a deformity reached half. A later study of forty-five children and adolescents with CMT and scoliosis examined how these curves behave and respond to treatment, and concluded that CMT scoliosis differs in important respects from ordinary adolescent scoliosis, with a marked tendency to an exaggerated forward curvature of the upper back. A large analysis of one hundred and seventy-five patients with hereditary motor and sensory neuropathy examined the deformities in detail and across genetic subtypes, reinforcing that spinal curvature is a genuine feature of the disease rather than a chance accompaniment.

Two themes recur across these studies. The first is that the deformity tends to appear during the growing years, often in early adolescence, with the average age at recognition in the region of twelve to thirteen. The second is that certain groups appear more susceptible, with female sex and the demyelinating type one form of CMT identified as carrying a higher likelihood of spinal involvement. These are tendencies described across populations, not predictions for any one person.

The neuromuscular mechanism: why a CMT curve is not an ordinary curve

To understand why CMT scoliosis is treated as its own entity, it helps to set it beside the far commoner adolescent idiopathic scoliosis, the curvature that arises in otherwise healthy teenagers for reasons that remain unknown. The two can look superficially similar on an X-ray, but they arise from different roots and tend to behave differently.

Adolescent idiopathic scoliosis occurs in a child whose nerves and muscles are entirely normal; the curve develops despite a balanced muscular framework, and the trunk muscles, though they cannot prevent the deformity, work properly. A neuromuscular curve, by contrast, develops precisely because the muscular framework is no longer balanced. In CMT the imbalance flows from the unreliable nerve supply to the trunk and the muscles that stabilise the spine and pelvis. The curve is, in a sense, a downstream consequence of the neuropathy.

Several practical differences follow from this. Neuromuscular curves, as a class, are described as having a greater tendency to progress, to extend over longer segments of the spine, and to be accompanied by an exaggerated kyphosis, the forward rounding of the upper back, which in CMT is a particularly noted feature. Because the underlying muscular weakness does not resolve, the forces that produced the curve persist, and this is part of why bracing, which can be helpful in idiopathic curves, is generally reported to be less successful in CMT. The deformity also sits within a wider pattern of musculoskeletal involvement, alongside the foot deformities and, in some, abnormalities of the hips, so that the spine is one part of a broader picture rather than an isolated finding. None of this is a statement about any individual; it is the general character of neuromuscular curvature as the literature describes it.

Kyphosis and the sagittal profile in CMT

When the spine is viewed from the side rather than from behind, it shows a series of gentle, alternating curves: a forward hollow in the neck and lower back, the lordosis, and a backward rounding in the chest region, the kyphosis. These curves, taken together, make up what is called the sagittal profile, and keeping them in proportion is part of how the body holds the head balanced economically over the pelvis. A recurring observation in CMT is that the curvature is frequently not confined to the sideways plane. An exaggerated rounding of the upper back, a thoracic hyperkyphosis, is described as a characteristic accompaniment of the scoliosis, so that the deformity is often a kyphoscoliosis rather than a pure sideways curve.

This sagittal dimension matters because it changes the character of the deformity and the way it is thought about. A combined curve places different demands on the trunk muscles, on balance when sitting and standing, and on the way the spine carries load. Where the muscles that would normally hold the upper back upright are weakened, the chest can drift forward into a stoop that the body then tries to compensate for elsewhere along the spine and at the pelvis. The recognition that CMT deformity has this strong forward component, rather than being a simple lateral bend, is one of the features that distinguishes it from ordinary adolescent scoliosis and one of the reasons it is followed and assessed in three dimensions rather than two.

The hips, the pelvis and the foundation of the spine

A curve of the spine is never quite a problem of the spine alone, because the spine sits upon the pelvis, and the pelvis in turn depends upon the hips. CMT is associated not only with spinal deformity but, in a proportion of patients, with dysplasia of the hips, a condition in which the ball of the hip joint is incompletely covered by its socket. A large study of children and adolescents with CMT found hip dysplasia in a substantial minority, with rates that depended on the age examined and the genetic subtype, and noted that a number of these hips required surgical attention. The same neuromuscular imbalance that can draw the spine into a curve appears, in some, to affect the developing hip joint as well.

This matters to the spinal picture because the pelvis is the platform on which the whole spinal column rests. If the pelvis is tilted, a state called pelvic obliquity, the spine above it must accommodate the slope, and a level, balanced pelvis is part of what allows a person to sit and stand comfortably with the head centred over the hips. In neuromuscular deformity generally, the relationship between the curve, the tilt of the pelvis and the position of the hips is considered together, because they form a single mechanical chain from the base of the skull to the feet. Viewing the spine, the pelvis and the hips as connected, rather than in isolation, is part of what makes the assessment of a neuromuscular curve distinct from that of an idiopathic one.

Genetics and subtypes, in relation to the spine

CMT is genetically diverse, with a long and growing list of responsible genes, but a few patterns are relevant to the spine. The commonest form by a wide margin is CMT type 1A, caused by a duplication of a small piece of chromosome seventeen that carries the gene for a myelin protein called PMP22. This duplication accounts for a large share of all CMT and underlies the demyelinating type one disease, in which the insulating sheath of the nerves is primarily affected. It is this type one group that has been associated with a higher likelihood of spinal deformity.

A particularly instructive example, from a spinal point of view, is CMT type 4C, a recessively inherited form caused by mutations in a gene called SH3TC2. In this subtype, scoliosis and kyphoscoliosis are so consistent a feature that a series describing the condition found spinal deformity in almost all affected individuals, and frequently as the presenting complaint, the very first sign that brought the person to attention. CMT4C illustrates, more starkly than the commoner subtypes, that in certain genetic forms the spine is not a peripheral concern but a central part of the clinical picture, and that a curve in a child can sometimes be the doorway to the diagnosis of the underlying neuropathy. The broad message is that the genetic subtype shapes the likelihood and the prominence of spinal involvement, with the demyelinating forms and CMT4C in particular standing out.

How spinal involvement in CMT is assessed

The assessment of a spinal curve in CMT follows the same broad principles as the assessment of any scoliosis, but it is carried out in the knowledge that the patient has an underlying neuropathy, and this knowledge shapes the emphasis. The starting point is clinical: the pattern of the curve, its flexibility, the balance of the trunk over the pelvis, the level of the shoulders and the position of the head, together with the wider neurological picture of the limbs, the feet and the hands.

Plain radiographs, taken with the person standing where possible, remain the foundation. They allow the curve to be measured by the standard Cobb angle, both in the sideways plane and from the side to capture the kyphosis, and serial films taken over time are the principal means of judging whether a curve is progressing during growth. Because the deformity in CMT so often has a forward component, the side view is given particular weight.

Magnetic resonance imaging, the MRI scan, is used where the spinal cord and nerve roots need to be examined, and to ensure that the curve is explained by the known neuromuscular process rather than by an additional structural problem within the spinal canal.

Assessment of the hips and pelvis, by examination and by imaging of the pelvis, is considered alongside the spine because of the recognised association with hip dysplasia and because the tilt of the pelvis influences the curve above it. Throughout, the curve is understood in three dimensions and as one element of a whole-body neuromuscular picture, rather than as an isolated measurement on a single film.

What the evidence shows about management

It is important to state at the outset that the evidence on managing spinal deformity in CMT rests on retrospective series and the broader literature on neuromuscular scoliosis, rather than on large randomised trials. The descriptions that follow report what these studies observe, not guidance for any person, and decisions in practice are individualised and made by the treating team.

Observation is the course described for curves that are small and not progressing, with monitoring through the growing years by serial examination and radiographs to detect any change. Because the curve in CMT can progress while a child is still growing, the literature emphasises that the period of most rapid growth is when watchfulness is most relevant.

Bracing, which has an established place in moderate idiopathic curves, is reported across the CMT literature to be of more limited value, because the persistent underlying muscular imbalance tends to work against it. Several series note that bracing in CMT scoliosis is often unsuccessful in halting progression, although it is sometimes described as a means of managing a curve through a period of growth rather than as a definitive solution.

Surgical correction, in the form of a spinal fusion with modern instrumentation, is described in the literature for curves that are large or progressive. Reported outcomes have been broadly encouraging, with substantial correction of the curve achieved and, notably, without the high rate of complications that might be feared in a patient with an underlying neuropathy. A particular and well recognised consideration is the difficulty of intraoperative monitoring of the nervous system: because CMT affects the peripheral nerves, the electrical signals used during surgery to check the spinal cord and nerve pathways can be hard to obtain, and older series reported success in only a minority of patients. More recent work has shown that with technical refinements, such as adjusting the settings used to evoke and record these signals, reliable monitoring can frequently be achieved in the CMT population, which is reassuring for the conduct of deformity surgery. Decisions on whether and when to operate, the levels involved, and whether the correction extends to the pelvis are weighed individually against the magnitude and progression of the curve and the wider condition of the patient.

In summary, the evidence points towards observation for small, stable curves, a cautious view of bracing given the underlying muscular imbalance, and reconstructive surgery considered for large or progressive deformity, all

within a framework of individualised, shared decision-making rather than fixed rules.

Surgical considerations in a neuropathic patient

Correcting a spinal deformity in someone with CMT brings together the general principles of neuromuscular scoliosis surgery with the particular features of an inherited peripheral neuropathy, and several threads from the literature are worth drawing together impersonally.

The first is the goal of surgery. In neuromuscular deformity the aim is not only to reduce the angle of the curve but to restore a balanced spine that sits squarely over a level pelvis, allowing comfortable, economical posture when sitting and standing. Because the deformity in CMT often combines a sideways curve with an exaggerated forward kyphosis, correction is considered in three dimensions, addressing both planes.

The second is the question of how far the correction should extend, and in particular whether it should reach down to the pelvis. In neuromuscular curves the decision to fuse to the pelvis is described in the literature as turning on the underlying disease and the behaviour of the curve and the pelvic tilt, rather than on the pelvic obliquity alone, and this is one of the recognised judgements in planning a long correction. The recognised association of CMT with hip dysplasia adds to the importance of considering the spine, the pelvis and the hips together as one mechanical chain.

The third, and most distinctive to CMT, is intraoperative neuromonitoring. During deformity surgery the function of the spinal cord and nerves is watched continuously using evoked electrical signals, so that any threat to the nervous system can be detected as it happens. In a disease that damages the peripheral nerves, these signals can be small and difficult to obtain from the outset, and the historical literature recorded that monitoring was frequently impossible in CMT. The more recent message is more optimistic: with deliberate technical adjustments the signals can often be recovered reliably, restoring this important safeguard. Set against these particular considerations, the broadly favourable complication profile reported in the published CMT surgical series is reassuring, though every operation carries its own recognised risks, which are weighed individually.

Outlook

The outlook for the spine in CMT is, for most people, a secondary part of a condition whose principal effects are felt in the feet and hands, and many people with CMT never develop a spinal curve at all. Where a deformity does arise, much depends on the same factors that govern neuromuscular curves more generally: the size of the curve, whether it is progressing, the stage of growth, the genetic subtype, and the wider neuromuscular condition of the person.

For small and stable curves the picture is one of monitoring rather than intervention, with many curves followed through growth without ever needing treatment. For larger or progressive deformity, the surgical literature offers a measure of reassurance, describing substantial and durable correction with a complication profile that has not proved as forbidding as the underlying neuropathy might suggest, and with the historical obstacle of intraoperative monitoring increasingly surmountable.

Across the whole field, the consistent themes are that CMT carries a genuinely raised likelihood of spinal curvature compared with the general population, that this curvature is neuromuscular in origin and therefore behaves differently from ordinary adolescent scoliosis, that it frequently combines a sideways curve with a forward kyphosis, and that the spine is best understood as one link in a chain that runs from the nerves of the limbs through the hips and pelvis to the curve itself. Any individual outlook is a matter for assessment by the treating team rather than something that can be read from a category or a measurement alone.