

Congenital Insensitivity to Pain and the Spine

A patient guide

In brief: Congenital insensitivity to pain with anhidrosis, known in the medical literature as hereditary sensory and autonomic neuropathy type IV, or HSAN IV, is a rare inherited disorder in which the nerves that carry pain and temperature sensation never develop properly. The cause is a fault in a single gene, *NTRK1*, which encodes the receptor through which nerve growth factor instructs certain sensory and autonomic nerve cells to survive and mature. Without that signal those nerves fail to form, and the affected person grows up unable to feel pain, unable to sense temperature, and unable to sweat. From the point of view of a spinal surgeon, the central problem is not the absence of pain itself but its consequence: pain is the body's alarm, and without it the spine is repeatedly injured without the person ever knowing. Small, unperceived insults accumulate. Fractures pass unnoticed and unprotected, joints between the vertebrae are loaded and damaged again and again, and a destructive, disintegrating arthropathy of the spine, a neuropathic or Charcot spine, can develop. Layered on top of this are a strong tendency to scoliosis and other deformity, and a surgical environment that is among the most unforgiving in all of spinal practice, with high rates of infection, failure of the bone to knit, hardware loosening and recurrent breakdown. The single most important idea is that the missing alarm, not a primary weakness of bone, is what allows the spine to be destroyed.

What congenital insensitivity to pain is, and why the spine is at risk

Congenital insensitivity to pain with anhidrosis is one of a small family of conditions, the hereditary sensory and autonomic neuropathies, in which the sensory and autonomic nervous systems are abnormal from birth. In the type designated HSAN IV the fault lies in the *NTRK1* gene, which carries the instructions for a receptor called TrkA. This receptor sits on the surface of particular nerve cells and receives a chemical messenger, nerve growth factor, that tells those cells to grow, mature and survive. The nerves that depend on this signal are the small fibres that carry pain and temperature sensation from the body to the spinal cord, and certain nerves of the autonomic system, including those that drive the sweat glands. When the receptor does not work, these nerves never properly form. The result is a person who cannot feel pain anywhere in the body, cannot sense heat or cold reliably, and cannot sweat, the feature termed anhidrosis.

It is worth being clear about what is preserved and what is lost. The sense of light touch, of vibration and of joint position is usually largely intact, because these travel by different, larger nerve fibres. What is missing is the protective sense, the sharp signal that says an action is harmful and must stop. Pain, uncomfortable as it is, is the mechanism by which a normal spine protects itself, the reason a person shifts position before a joint is overloaded, guards a strained back, rests a healing fracture and seeks help when something is seriously wrong. Remove that signal and the spine loses its capacity for self-protection.

This is why the spine is so vulnerable in this condition, and why a spinal surgeon needs to understand it. The bone itself is not primarily abnormal; rather, it is repeatedly subjected to forces that a person with normal sensation would instinctively avoid. The damage is mechanical and cumulative, the product of countless small injuries that are never felt, never guarded and never allowed to heal. Understanding congenital insensitivity to pain through the lens of the spine therefore means understanding the consequences of a silent, unprotected back rather than the biology of pain in the abstract.

The neuropathic, or Charcot, spine

The most characteristic and serious spinal manifestation of congenital insensitivity to pain is a destructive arthropathy known as a neuropathic spine, or Charcot spine, named after the same process of joint disintegration

first described in insensate limbs. It is the spinal expression of a principle seen throughout the body in conditions that abolish protective sensation: a joint that cannot feel pain is a joint that is destroyed by its own use.

The mechanism is one of repetitive, unperceived microtrauma. Each movement and load that a normal spine would moderate through the discomfort it produces is, in the insensate spine, taken to and beyond its limit without restraint. The small joints and the discs, and the bone of the vertebrae themselves, are subjected again and again to forces that would ordinarily be guarded against. Over months and years the cartilage wears, the bone fragments, the discs collapse, and the architecture of one or more spinal segments breaks down. The body's attempt at repair is disordered, producing new bone, debris and fluid around a joint that continues to be loaded, so that the segment becomes simultaneously destroyed and grossly disorganised.

The radiological picture is often dramatic and out of all proportion to the symptoms. Because the person feels little or nothing, a vertebral segment may be largely destroyed, with collapse, fragmentation, sclerosis and abnormal new bone, before the problem declares itself. It frequently comes to attention not through pain but through a deformity that becomes visible, a swelling, an audible or palpable clunking as the unstable segment moves, a loss of height or function, or, in the most serious cases, the gradual development of pressure on the spinal cord or nerves. The published case literature repeatedly describes patients in whom a single segment, often in the lower back, has disintegrated to a degree that would be agonising in anyone with normal sensation, yet which has been tolerated and therefore allowed to progress.

Fractures and vertebral injury that pass unnoticed

A separate but related theme is the unnoticed fracture. In the general population a vertebral fracture announces itself with pain, and that pain is what causes a person to rest, to protect the back and to seek assessment, all of which give the fracture the chance to heal in a good position. In congenital insensitivity to pain none of that happens. A fracture of a vertebra may occur during ordinary activity, or after a minor stumble, and produce no signal at all. The person continues to move, to bear weight and to load the injured bone exactly as before.

The consequences flow from this lack of protection. A fracture that is never rested is a fracture that continues to be displaced, that may not heal, or that heals in a deformed position. Repeated loading of an unhealed vertebral injury can merge with the neuropathic process described above, so that what begins as a single unnoticed fracture becomes the seed of a wider segmental collapse and destruction. The orthopaedic literature on this condition, drawn largely from case series, records high rates of fractures across the skeleton precisely because injuries are not felt and therefore not protected, and the spine is not exempt from this pattern.

There is a further difficulty in detection. Because the usual presenting complaint, pain, is absent, the timeline of injury is often unknown. A spine may show evidence of old, healed and ongoing injury all at once, with no clear history of a single event, because each individual insult was silent. This makes the spine in congenital insensitivity to pain a record of accumulated, undated trauma rather than of any one identifiable accident.

Scoliosis and spinal deformity

Deformity of the spine is common in congenital insensitivity to pain and in the closely related sensory and autonomic neuropathies. Scoliosis, a sideways curvature with rotation, and kyphosis, an excessive forward bend, are both described, and they arise through more than one route.

In part, deformity is the visible end result of the destructive and fracturing processes already described: when segments collapse, fragment or heal in a deformed position, the overall alignment of the spine is thrown out. In part, it reflects the loss of the normal, sensation-guided muscular control that keeps the growing spine balanced, so that a curve, once begun, is not corrected by the protective adjustments a normally innervated spine would make. The allied condition familial dysautonomia, another of the hereditary sensory and autonomic neuropathies, is well documented as causing scoliosis and kyphosis in a very high proportion of those affected, and provides a useful model for understanding how a spine deprived of normal sensory feedback tends towards progressive deformity.

What sets deformity in this condition apart is its tendency to progress in the setting of insensate, fragile bone and disordered healing, and the fact that the usual safeguards against worsening are absent. A curve that would be guarded, braced and monitored against a background of normal sensation here progresses in a spine that gives no warning, which is one reason these deformities can become severe before they are addressed.

Why surgery on this spine is so difficult

It is in the operating theatre that congenital insensitivity to pain reveals itself as one of the most challenging conditions a spinal surgeon can encounter. The very features that destroy the spine in the first place also conspire to undermine its surgical reconstruction, and the case literature is consistent in describing high rates of complication, repeated failure of fixation and the frequent need for revision.

The first difficulty is the bone and the segment itself. The neuropathic, Charcot process leaves bone that is fragmented, disorganised and abnormally remodelled, a poor foundation into which to anchor screws and across which to achieve a solid fusion. The destructive process that necessitated surgery does not switch off because an operation has been performed; the same insensate loading that destroyed the segment continues to act on the reconstruction.

The second difficulty is healing. Achieving a fusion, the knitting of bone across a segment into a solid block, depends on a biological environment that, in this condition, appears repeatedly to be impaired. The case series describe high rates of non-union, or pseudarthrosis, where the bone simply fails to bridge. Where fusion does not take, the metalwork is left to carry load indefinitely and eventually loosens, breaks or pulls out of the bone, a sequence documented again and again in reports of repeated hardware failure, migration and the need for multiple revision operations, sometimes extending the construct ever further along the spine.

The third difficulty is infection. These patients are prone to wound problems and to deep infection, a tendency rooted both in the autonomic and skin abnormalities of the condition, including the absence of sweating and impaired skin regulation, and in the difficulty of detecting an infection that produces no pain. The orthopaedic literature on spinal deformity correction in this group specifically notes high rates of infection and reoperation, and the broader surgical experience records recurrent osteomyelitis and impaired control of infection.

The fourth difficulty is the cycle of recurrent breakdown. Because the underlying driver, the insensate, unprotected loading of the spine, persists after any operation, reconstructions are at constant risk of breaking down again at the same or an adjacent level, so that surgery in this condition is frequently not a single definitive event but a sequence of interventions over years. The long-term follow-up reports in the literature describe exactly this: recurrent collapse, repeated revision and the steady extension of fusions in an attempt to keep pace with ongoing destruction.

How it is assessed

Assessment of the spine in congenital insensitivity to pain rests heavily on imaging, precisely because the usual clinical signal of pain is absent and cannot be relied upon to indicate the presence or severity of a problem.

Plain X-rays, including standing and bending views, are the foundation, used to document deformity, to measure curvature and alignment, and to reveal the destruction, collapse, fragmentation and abnormal new bone that mark a neuropathic segment. Because the condition is one of accumulated, undated injury, comparison of images over time is particularly valuable in showing whether a segment is stable or progressing.

Computed tomography, the CT scan, gives the detailed bony picture needed to understand the extent of destruction, the quality of the bone available for fixation and the state of any previous fusion or hardware. Magnetic resonance imaging, the MRI scan, is used to assess the soft tissues, the discs, and above all whether the spinal cord or nerves are being compressed by an unstable or collapsing segment, a question of real importance given that the person may not feel the warning signs of neural compression. Because infection is both common and difficult to detect by symptoms in this group, imaging and laboratory assessment also carry a particular weight

in identifying it.

Throughout, the recurring theme is that the spine in this condition cannot be assessed in the way most spines are, through the story of a person's pain, and that objective imaging therefore takes on a heightened role.

What the evidence shows about management

It must be said plainly that the evidence base for managing the spine in congenital insensitivity to pain is built almost entirely on individual case reports, small case series and reviews of those reports. This is a rare condition, and there are no trials, no agreed protocols and no large comparative studies to define what should be done. What follows describes what the published experience reports, not guidance for any individual, and decisions in practice are highly individualised.

The literature describes a tension at the heart of management. On one hand, a destroyed, unstable or deforming spinal segment, particularly one threatening the spinal cord or nerves, may appear to demand surgical stabilisation. On the other, the same reports document just how poorly this spine tolerates surgery, with the high rates of non-union, infection, hardware failure and recurrent breakdown already described. The published experience therefore reflects a difficult balance rather than a clear path, with some reports describing prolonged attempts at non-operative management and protection, and others describing extensive reconstructive surgery, often three-column or circumferential fusion, undertaken when instability or neural compromise leaves little alternative.

Where surgery is described, the principles that emerge from the case literature are those of robust, often extensive fixation across the destroyed segment, attention to achieving fusion in a hostile biological environment, vigilance for infection, and an expectation that revision may be needed. Several long-term reports document the reality that a single operation rarely settles the matter and that ongoing destruction may require staged or repeated intervention. Prevention of further injury through protection and monitoring is a recurring theme on the conservative side, reflecting the understanding that the root problem, the absent protective signal, cannot itself be restored.

In summary, the published evidence points to a condition in which both inaction and surgery carry serious difficulties, in which management is necessarily tailored to the individual segment, the stability of the spine and the state of the neural structures, and in which decisions are made within a framework of individualised, shared assessment by the treating team rather than according to any fixed rule.

Outlook

The outlook for the spine in congenital insensitivity to pain is shaped, above all, by the fact that the underlying cause is permanent. The genetic fault that abolishes protective sensation does not change, and so the tendency to unperceived injury, destruction and deformity persists throughout life. This is a chronic condition of accumulated damage rather than a single event from which one recovers.

That said, the picture is not uniformly bleak, and a great deal depends on the circumstances of the individual. Much rests on how early the spine's vulnerability is recognised, on whether deformity and destruction can be limited before they become severe, on the stability of the spine and whether the neural structures are threatened, and, where surgery is undertaken, on whether a fusion can be achieved and held in a spine that resists healing and continues to be loaded. The long-term reports in the literature describe both patients who have stabilised after reconstruction and those who have required repeated revision over many years, and the difference between these courses lies in this combination of factors rather than in any single one.

Across the whole field the consistent themes are these: that the absence of pain, not a primary disease of bone, is what places the spine in jeopardy; that injury and destruction proceed silently and may be advanced before they are noticed; that deformity is common and tends to progress; that surgery is uniquely difficult and prone to complication and recurrence; and that any individual outlook is a matter for careful, ongoing assessment by the treating team rather than something that can be read from the diagnosis alone.