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Inborn analgesia

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Date of submission: 28/6/2018

Abstract:

Inborn analgesia also known as congenital insensitivity to pain is a condition, present from birth, that inhibits the ability to perceive physical pain. Affected individuals are unable to feel pain in any part of their body. Over time, this lack of pain awareness can lead to an accumulation of injuries and health issues that may affect life expectancy. Here in this report the way of taking care of those people will be discussed(1).

Introduction:

Pain is a sensory modality present in all complex organisms and used to detect potential and real tissue damage. Although it is an unpleasant sensory and emotional experience, it substantially affects our behaviour, providing a survival advantage. Normally, pain is detected by a complex system of mechanical and chemical sensors called nociceptors, which then send information through spinal interneuronal pathways to the brain. However, in a number of rare conditions, components of the pain-signalling pathway may be impaired or fail to develop. One such condition is congenital insensitivity to pain (CIP), where individuals are unable to perceive pain from birth.There are two common forms of CIP. First, loss-of-function mutation in the *SCN9A* renders nociceptors unable to respond to any noxious stimulus. Second, loss-of-function mutation in the *NTRK1* leads to a failure of nociceptors to develop. Recently, a new form of CIP has been described, caused by mutations in an epigenetic regulator *PRDM12*. The phenotype of these patients was briefly annotated in the initial study(2).

Discussion:

Congenital insensitivity to pain is a condition that inhibits the ability to perceive physical pain. Affected individuals never feel pain in any part of their body when injured. People with this condition can feel the difference between sharp and dull and hot and cold, but cannot sense, for example, that a hot beverage is burning their tongue. This lack of pain awareness often leads to an accumulation of wounds, bruises, broken bones, and other health issues that may go undetected. Young children with congenital insensitivity to pain may have mouth or finger wounds due to repeated self-biting and may also experience multiple burn-related injuries. These repeated injuries often lead to a reduced life expectancy in people with congenital insensitivity to pain. Many people with congenital insensitivity to pain also have a complete loss of the sense of smell (anosmia).Congenital insensitivity to pain is considered a form of peripheral neuropathy because it affects the peripheral nervous system, which connects the brain and spinal cord to muscles and to cells that detect sensations such as touch, smell, and pain.

Mutations in the *SCN9A* gene cause congenital insensitivity to pain. The *SCN9A* gene provides instructions for making one part (the alpha subunit) of a sodium channel called NaV1.7. Sodium channels transport positively charged sodium atoms (sodium ions) into cells and play a key role in a cell's ability to generate and transmit electrical signals. NaV1.7 sodium channels are found in nerve cells called nociceptors that transmit pain signals to the spinal cord and brain. The NaV1.7 channel is also found in olfactory sensory neurons, which are nerve cells in the nasal cavity that transmit smell-related signals to the brain(3).

The *SCN9A* gene mutations that cause congenital insensitivity to pain result in the production of nonfunctional alpha subunits that cannot be incorporated into NaV1.7 channels. As a result, the channels cannot be formed. The absence of NaV1.7 channels impairs the transmission of pain signals from the site of injury to the brain, causing those affected to be insensitive to pain. Loss of this channel in olfactory sensory neurons likely impairs the transmission of smell-related signals to the brain, leading to anosmia. This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition. Congenital insensitivity to pain cannot be cured. Because affected people do not feel pain, preventing injuries and finding injuries quickly is important. To encourage prompt detection of injuries, patient/parents should conduct daily self-checks particularly of vulnerable regions such as their feet, hands and joints.

Doctors should look for broken bones, even if the patient feels no pain. Performing X-rays and other imaging tests at sites of possible injury is recommended. An annual skeletal MRI may also be recommended. Patients are at increased risk of repeated Staphylococcal infections that arise without an obvious external cause, see Results. The early use of topical antibacterial creams until all signs of inflammation or infection have resolved was reported to be highly effective in preventing chronic infection. For more significant infections, the lack of pain as a sign had hindered the diagnosis of osteomyelitis and septic arthritis, and all involved physicians should be aware of this. Most significant infections were complicated and required a course of IV antibiotics before they resolved. The diagnostic criteria generally agreed upon for this disorder include a generalized indifference to pain dating from birth; no impairment of other sensory modalities; normal intelligence; normal deep tendon reflexes; no visceral pain perception; normal skin biopsy; no diminution of myelinated or un-myelinated nerve fibres in sural nerve biopsy; normal motor and sensory nerve conduction velocities; and normal karyotype(4).

Interestingly, those with congenital analgesia can still feel sensations such as normal body-to-body contact, which means that the brain can receive some information filtered through the nervous system: Perception of passive movement, joint position, and vibration is normal, as are tactile thresholds and light touch perception. Likewise, the ability to distinguish sharp and dull stimuli and detect differences in temperature also appears to remain intact. Reflexes are maintained. However, when it comes to extreme temperature changes, or any bodily damage that signals the body to react in an emergency fashion, the body simply doesn't respond.

Conclusion:

As patients with CIP are unable to feel pain, this impairs their ability to detect injuries promptly and appropriately immobilise the injury to allow for healing. To encourage prompt detection of injuries, patient/parents should conduct daily self-checks particularly of vulnerable regions such as their feet, hands and joints. The medical management of any injuries is as for other forms of CIP.

References:

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