Congenital Insensitivity to pain

 Congenital insensitivity to pain and anhidrosis (CIPA) is a rare recessive genetic disorder in which people cannot feel pain and temperature and suffers from decreased or absent sweating. This condition is also known as hereditary sensory and autonomic neuropathy type IV. The signs and symptoms of CIPA appear early, usually at birth or during infancy, but with careful medical attention, affected individuals can live into adulthood. An inability to feel pain and temperature often leads to repeated severe injuries. Unintentional self-injury is common in people with CIPA, typically by biting the tongue, lips, or fingers, which may lead to spontaneous amputation of the affected area. (Service) In addition, people with CIPA heal slowly from skin and bone injuries. Repeated trauma can lead to chronic bone infections (osteomyelitis) or a condition called Charcot joints, in which the bones and tissue surrounding joints are destroyed. Normally, sweating helps cool the body temperature. However, in people with CIPA, anhidrosis often causes recurrent, extremely high fevers (hyperpyrexia) and seizures brought on by high temperature (febrile seizures). In addition to the characteristic features, there are other signs and symptoms of CIPA. Many affected individuals have thick, leathery skin (lichenification) on the palms of their hands or misshapen fingernails or toenails. They can also have patches on their scalp where hair does not grow (hypotrichosis). About half of people with CIPA show signs of hyperactivity or emotional instability, and many affected individuals have intellectual disability. Some people with CIPA have weak muscle tone (hypotonia) when they are young, but muscle strength and tone become more normal as they get older. Mutations in the *NTRK1* gene cause CIPA. The *NTRK1* gene provides instructions for making a receptor protein that attaches (binds) to another protein called NGFβ. The NTRK1 receptor is important for the survival of nerve cells (neurons). Mutations in the *NTRK1* gene lead to a protein that cannot transmit signals. Without the proper signaling, neurons die by a process of self-destruction called apoptosis. Loss of sensory neurons leads to the inability to feel pain in people with CIPA. In addition, people with CIPA lose the nerves leading to their sweat glands, which cause the anhidrosis seen in affected individuals.

 Recently, researchers have successfully generated a sense of touch and pressure through electrical signals. This can potentially help people with prosthetics and people with CIPA. The first set of experiments aimed to develop an algorithm to determine the location of a touch. First, researchers taught monkeys how to react in a certain way when they felt something touch them on each finger. Then they mapped the brain activity that occurred when the monkeys’ fingers were touched and placed electrodes at the locations that were activated. An electrical stimuli was applied to the electrodes and the monkeys reacted in the same way like they did when they were physically touched (Service). After showing that they could successfully map touch location through an electric stimulus, researchers determined how to implement the sense of pressure. They developed an algorithm that created a specified magnitude of electric current that made a sensation of pressure. Just like in the previous experiments, the monkeys responded the same with and without the physical touch. There are two basic kinds of touch; fine touch and crude touch. Fine touch allows for localization of the touch whereas crude touch allows for recognition of touch but inability to decipher where exactly the touch came from (Service). Through these experiments, the researchers were successfully able to implement fine touch into electric signals. This discovery can help people with CIPA because they now might be able to experience pain, which plays a key survival role by warning about potential or actual injuries. Without the sensation of pain, people suffering from CIPA can harm themselves repeatedly without even realizing it. Therefore, a cure is needed to provide aid for these people, which these researchers are working on.

Sources

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