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INSENSITIVITY TO PAIN WITH ANHIDROSIS-A MULTISYSTEMIC DISORDER

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Congenital Insensitivity to Pain with Anhydrosis (CIPA) is a rare autosomal recessive inherited disorder, cause by a mutation in the TRKA gene encoding for the specific NGF receptor. The disease is characterized by lack of reaction to noxious stimuli, recurrent episodes of unexplained fever (in infancy), anhydrosis, self mutilative behavior and mental retardation.
In the Bedouin population of the Negev (southern part of Israel) this syndrome is quite prevalent, probably due to increased incidence intermarriage.
37 patients with the syndrome are treated in our center. Twenty eight of them carry the 1926 ins T mutation in the TRKA gene.
All patients demonstrate a multi systemic involvement including: mental retardation, self mutilation to finger tips, tongue and eyes, recurrent bone trauma, osteomyelitis, susceptibility to infection and significantly delayed wound healing. Of particular concern are disorders of dentition, tongue mutilation and jaw osteomyelitis following teeth extraction or local trauma.
Endocrine involvement, autonomic changes and immunodeficiency are also associated.
The treatment of these patients is complex and involves a multi disciplinary team including general pediatrician, orthopedic, ophthalmic and plastic surgeons, social worker and pediatric neurologist as well as highly qualified nursing personnel in purpose to maintain an optimal comprehensive care of these complex patients.