

A Case of Congenital Insensitivity to Pain With Anhidrosis Comorbid With Attention Deficit Hyperactivity Disorder: Clinical Implications for Pathophysiology and Treatment.

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J Nerv Ment Dis. 2018 Apr;206(4):296-299.
doi: 10.1097/NMD.0000000000000785.

Abstract

Congenital insensitivity to pain with anhidrosis (CIPA) is a rare autosomal recessive genetic disorder caused by a mutation in the neurotrophic tyrosine kinase receptor (NTRK1) gene. CIPA is accompanied by abnormal catecholamine metabolism and decreased blood concentration of dopamine and norepinephrine. Attention deficit hyperactivity disorder (ADHD) is a neurodevelopmental disorder of heterogeneous etiology and presentation, and recent reports have suggested a pathophysiological role of neurotrophins in ADHD. Furthermore, dopamine and norepinephrine are known to play major roles in the pathophysiology of ADHD, and the imbalance of monoaminergic and cholinergic systems as an underlying cause of ADHD has recently been studied. Here, we report the case of an 11-year-old boy with CIPA and comorbid ADHD. Our observations have important clinical implications for patients with CIPA. Because of deficiencies in self-control, proper management of these patients necessitates a highly structured and monitored environment, made dually important by possible comorbidity of ADHD.