A newly identified chromosomal microdeletion and an N-box mutation of the AChR epsilon gene cause a congenital myasthenic syndrome

ID 155367
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Year 2002
Title A newly identified chromosomal microdeletion and an N-box mutation of the AChR epsilon gene cause a congenital myasthenic syndrome
Journal Brain
Volume 125
Number 5
Pages / **Article-Number** 1005-1013
Keywords acetylcholine receptor, congenital myasthenic syndrome, epsilon subunit, gene deletion, promoter mutation
Congenital myasthenic syndromes (CMSs) are frequently caused by mutations of the coding region of the acetylcholine receptor epsilon subunit (AChRepsilon) gene leading to a reduced expression of the acetylcholine receptor (AChR) at the postsynaptic membrane. Two recent observations have linked two different N-box mutations of the human AChRepsilon promoter to a clinical CMS phenotype. N-boxes are regulatory sequence elements of mammalian promoters that confer synapse-specific expression of several genes, including the AChR subunit genes. Here, we report on a novel point mutation (epsilon-154G–
Publisher Oxford University Press
ISSN/ISBN 0006-8950 ; 1460-2156
edoc-URL http://edoc.unibas.ch/dok/A5258401
Full Text on edoc Available;
Digital Object Identifier DOI 10.1093/brain/awf095
ISI-Number WOS:000175626900009
Document type (ISI) Article