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The 2018 version of the gene table of monogenic neuromuscular disorders (nuclear genome)

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General features

This table is published annually in the December issue. Its purpose is to provide the reader of *Neuromuscular Disorders* with an updated list of monogenic muscle diseases due to a primary defect residing in the nuclear genome. It comprises diseases in which the causative gene is known or at least localized on a chromosome, if not yet identified. Diseases for which the locus has not been mapped or which are due to defects involving mitochondrial genes are not included.[1]

As in past years the diseases are classified into 16 groups:

1. *Muscular dystrophies;*
2. *Congenital muscular dystrophies;*
3. *Congenital myopathies;*
4. *Distal myopathies;*
5. *Other myopathies;*
6. *Myotonic syndromes;*
7. *Ion channel muscle diseases;*
8. *Malignant hyperthermias;*
9. *Metabolic myopathies;*
10. *Hereditary cardiomyopathies, subdivided into*
    10A *(non-arrhythmogenic) and*
    10B *(arrhythmogenic);*
11. *Congenital myasthenic syndromes;*
12. *Motor neurone diseases;*
13. *Hereditary ataxias;*
14. *Hereditary motor and sensory neuropathies;*
15. *Hereditary paraplegias;*
16. *Other neuromuscular disorders.*

In each group every entry corresponds to a clinical entity and has an item number.[2] A given gene may be involved in several different clinical entities (phenotypic heterogeneity such as in *LMNA* defects) and conversely a given clinical entity may be produced by a defect in several possible alternative genes (genotypic heterogeneity such as in CMT). In some diseases both kinds of heterogeneity may occur. As a consequence a gene or a disease may be cited in several places of the table.

The two versions of the gene table[3]

The **annual printed version** below is abridged and does not contain the *Arrhythmogenic Hereditary Cardiomyopathies (Group 10-B), Hereditary Ataxias (Group 13), and Hereditary Paraplegias (Group 15)*. The list of references is restricted to new key references corresponding to the items added or implemented since the preceding year.

The **full online version** contains the complete data of the 16 groups and the cumulative list of key references since 1991. It is freely accessible at http://www.musclegenetable.fr. It is designed to cope with the complexity described above. In this version the data are cross-referenced and linked to *PubMed* and to major databases related to molecular medicine *(Leiden Muscular Dystrophy, OMIM, NCBI, Genatlas, Orphanet, GeneCards)*. It contains several query tools allowing one to perform a variety of interrogations. This computerized version of the table is now surpassing the printed version which cannot accommodate the ever increasing volume and complexity of data. The **statistics tool** instantly provides the latest list of genes, proteins, phenotypes and cumulative bibliographic key references. Each list can be displayed, printed and exported in Excel format.

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[1] For diseases caused by mitochondrial genome mutations see: MITOMAP A human mitochondrial genome database. A compendium of polymorphisms and mutations of the human mitochondrial DNA http://www.mitomap.org/ MITOMAP.

[2] The assigned item number is provisional and may change in the next annual version.

[3] The history and development of both versions of the table are presented in the 2013 publication *(Kaplan JC and Hamroun D. The 2013 version of the gene table of neuromuscular disorders. Neuromuscul Disord. 22 (12), 1108–1135.)*

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Overview of the new data in the 2018 printed version of the gene table (pages 895 to 929 of this issue)

There are 35 new items, marked by background shading. Altogether they comprise **28 additional genes** and **8 additional phenotypic variants** caused by a gene already listed in the 2017 version (see box). The new key references of the printed version of the table are listed in pages 927–929 in this issue.

New in the 2017 printed version of the gene table
28 genes added:

AIFM1 (item # 16.34)
CALR3 (item # 10.20)
CNTNAP1 (item # 16.15)
DSG2 (item # 10.59)
ERBB4 (item # 12.57)
FASTKD2 (item # 16.36)
FLAD1 (item # 9.30)
HRAS (item # 3.50)
INPP5K (item # 2.49)
LAMA5 (item # 11.35)
MYMK (item # 3.51)
MYO18B (item # 3.49)
MYPN (item # 3.11 and item # 10.23)
NEFH (item # 14.66)
PMP2 (item # 14. 11)
POGLUT1 (item # 1.46)
PRDM1 (item # 10.68)
PTRH2 (item # 16.37)
RAF1 (item # 10.70)
SGPL1 (item # 14.80)
SLC18A3 (item # 11.27)
SLC25A46 (item # 12.73)
SPTBN4 (item # 3.47)
TMEM65 (item # 16.35)
TUBA4A (item # 12.60)
VAMP1 (item # 11.34 and item #15.66)
WARS (item # 12.23)
ZAK (item # 3.48)

8 additional phenotypic variants caused by mutation in a gene already listed in the gene table

SMCHD1
(item # 1.11 allelic to BAMS (OMIM #603457))
DAG1 (item # 2.37)
GYG1 (item # 9.12)
LDB3 (item # 10.25)
TCAP (item # 10.26)
BAG3 (item # 10.64 and 14.67)
VCP (item # 14.61)

1 new gene for a previously identified locus

SIGMAR1 (item #12.6, allelic to item # 12.54)

40 new key references

Citation of the gene table

- Printed version: Bonne G, F Rivier, Hamroun D. The 2018 version of the gene table of neuromuscular disorders. Neuromuscul Disord. 26 (12), 895–929.
- Online version: GeneTable of Neuromuscular Disorders: http://www.musclegenetable.fr

Contact

Users of the gene table are kindly requested to send any comments on the printed and/or the online version to g.bonne@institut-myologie.org.

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