A locus-specific database for mutations in GDAP1 allows analysis of genotype-phenotype correlations in Charcot-Marie-Tooth diseases type 4A and 2K

Titre	A locus-specific database for mutations in GDAP1 allows analysis of genotype-phenotype correlations in Charcot-Marie-Tooth diseases type 4A and 2K
Type de publication	Article de revue
Auteur	Cassereau, Julien [1], Chevrollier, Arnaud [2], Bonneau, Dominique [3], Verny, Christophe [4], Procaccio, Vincent [5], Reynier, Pascal [6], Ferré, Marc [7]
Editeur	BioMed Central
Type	Article scientifique dans une revue à comité de lecture
Année	2011
Langue	Anglais
Date	2011
Volume	6
Titre de la revue	Orphanet Journal of Rare Diseases
ISSN	1750-1172
Mots-clés	Charcot-Marie-Tooth Disease/genetics/physiopathology [8], Databases, Genetic [9], Electrophysiology [10], Female [11], Genes, Dominant [12], Genes, Recessive [13], Genetic Association Studies [14], Genetic Predisposition to Disease [15], Humans [16], Male [17], Mobility Limitation [18], Mutation [19], Nerve Tissue Proteins/genetics [20], Wheelchairs/utilization [21]
Résumé en anglais	BACKGROUND: The ganglioside-induced differentiation-associated protein 1 gene (GDAP1), which is involved in the Charcot-Marie-Tooth disease (CMT), the most commonly inherited peripheral neuropathy, encodes a protein anchored to the mitochondrial outer membrane. The phenotypic presentations of patients carrying GDAP1 mutations are heterogeneous, making it difficult to determine genotype-phenotype correlations, since the majority of the mutations have been found in only a few unrelated patients. Locus-specific databases (LSDB) established in the framework of the Human Variome Project provide powerful tools for the investigation of such rare diseases. METHODS AND RESULTS: We report the development of a publicly accessible LSDB for the GDAP1 gene. The GDAP1 LSDB has adopted the Leiden Open-source Variation Database (LOVD) software platform. This database, which now contains 57 unique variants reported in 179 cases of CMT, offers a detailed description of the molecular, clinical and electrophysiological data of the patients. The usefulness of the GDAP1 database is illustrated by the finding that GDAP1 mutations lead to primary axonal damage in CMT, with secondary demyelination in the more severe cases of the disease. CONCLUSION: Findings of this nature should lead to a better understanding of the pathophysiology of CMT. Finally, the GDAP1 LSDB, which is part of the mitodyn.org portal of databases of genes incriminated in disorders involving mitochondrial dynamics and bioenergetics, should yield new insights into mitochondrial diseases.

URL de la notice	http://okina.univ-angers.fr/publications/ua8159 [22]
DOI	10.1186/1750-1172-6-87 [23]
Lien vers le document	http://dx.doi.org/10.1186/1750-1172-6-87 [23]
Titre abrégé	Orphanet J Rare Dis

Liens
[1] http://okina.univ-angers.fr/julien.cassereau/publications
[2] http://okina.univ-angers.fr/arnaud.chevrollier/publications
[3] http://okina.univ-angers.fr/d.bonneau/publications
[4] http://okina.univ-angers.fr/ch.verny/publications
[5] http://okina.univ-angers.fr/v.procaccio/publications
[6] http://okina.univ-angers.fr/pascal.reynier/publications
[7] http://okina.univ-angers.fr/marc.ferre/publications
[8] http://okina.univ-angers.fr/publications?f[keyword]=12762
[9] http://okina.univ-angers.fr/publications?f[keyword]=12763
[10] http://okina.univ-angers.fr/publications?f[keyword]=12765
[11] http://okina.univ-angers.fr/publications?f[keyword]=1075
[12] http://okina.univ-angers.fr/publications?f[keyword]=12766
[13] http://okina.univ-angers.fr/publications?f[keyword]=12767
[14] http://okina.univ-angers.fr/publications?f[keyword]=12764
[15] http://okina.univ-angers.fr/publications?f[keyword]=11526
[16] http://okina.univ-angers.fr/publications?f[keyword]=991
[17] http://okina.univ-angers.fr/publications?f[keyword]=968
[18] http://okina.univ-angers.fr/publications?f[keyword]=12755
[19] http://okina.univ-angers.fr/publications?f[keyword]=1133
[20] http://okina.univ-angers.fr/publications?f[keyword]=12768
[21] http://okina.univ-angers.fr/publications?f[keyword]=12769
[22] http://okina.univ-angers.fr/publications/ua8159
[23] http://dx.doi.org/10.1186/1750-1172-6-87