Resource Summary Report

Generated by dkNET on Apr 16, 2025

DMLE

RRID:SCR_013454 Type: Tool

Proper Citation

DMLE (RRID:SCR_013454)

Resource Information

URL: http://www.dmle.org/

Proper Citation: DMLE (RRID:SCR_013454)

Description: Software application for high-resolution mapping of the position of a disease mutation relative to a set of genetic markers using population linkage disequilibrium (LD). (entry from Genetic Analysis Software)

Abbreviations: DMLE

Synonyms: Disease Mapping using Linkage disEquilibrium

Resource Type: software resource, software application

Keywords: gene, genetic, genomic, linux, ms-windows

Funding:

Resource Name: DMLE

Resource ID: SCR_013454

Alternate IDs: nlx_154218

Record Creation Time: 20220129T080316+0000

Record Last Update: 20250416T063642+0000

Ratings and Alerts

No rating or validation information has been found for DMLE.

No alerts have been found for DMLE.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 20 mentions in open access literature.

Listed below are recent publications. The full list is available at <u>dkNET</u>.

Islam S, et al. (2021) Founder mutation in the PMM2 promotor causes hyperinsulinemic hypoglycaemia/polycystic kidney disease (HIPKD). Molecular genetics & genomic medicine, 9(12), e1674.

Luo X, et al. (2021) Characterization of a Cohort of Patients With LIG4 Deficiency Reveals the Founder Effect of p.R278L, Unique to the Chinese Population. Frontiers in immunology, 12, 695993.

Esperón-Moldes U, et al. (2020) Novel CYP4F22 mutations associated with autosomal recessive congenital ichthyosis (ARCI). Study of the CYP4F22 c.1303C>T founder mutation. PloS one, 15(2), e0229025.

Zytsar MV, et al. (2020) High Rates of Three Common GJB2 Mutations c.516G>C, c.-23+1G>A, c.235delC in Deaf Patients from Southern Siberia Are Due to the Founder Effect. Genes, 11(7).

Kim YM, et al. (2020) The GBA p.G85E mutation in Korean patients with non-neuronopathic Gaucher disease: founder and neuroprotective effects. Orphanet journal of rare diseases, 15(1), 318.

Tatour Y, et al. (2019) A novel intronic mutation of PDE6B is a major cause of autosomal recessive retinitis pigmentosa among Caucasus Jews. Molecular vision, 25, 155.

Garagiola I, et al. (2016) A recurrent F8 mutation (c.6046C>T) causing hemophilia A in 8% of northern Italian patients: evidence for a founder effect. Molecular genetics & genomic medicine, 4(2), 152.

Caleca L, et al. (2014) Characterization of an Italian founder mutation in the RING-finger domain of BRCA1. PloS one, 9(2), e86924.

Winbo A, et al. (2014) Phenotype, origin and estimated prevalence of a common long QT syndrome mutation: a clinical, genealogical and molecular genetics study including Swedish

R518X/KCNQ1 families. BMC cardiovascular disorders, 14, 22.

de Alencar DO, et al. (2014) Fabry disease: Evidence for a regional founder effect of the GLA gene mutation 30delG in Brazilian patients. Molecular genetics and metabolism reports, 1, 414.

García-Murias M, et al. (2012) 'Costa da Morte' ataxia is spinocerebellar ataxia 36: clinical and genetic characterization. Brain : a journal of neurology, 135(Pt 5), 1423.

Liskova P, et al. (2012) High prevalence of posterior polymorphous corneal dystrophy in the Czech Republic; linkage disequilibrium mapping and dating an ancestral mutation. PloS one, 7(9), e45495.

Hünemeier T, et al. (2012) Evolutionary responses to a constructed niche: ancient Mesoamericans as a model of gene-culture coevolution. PloS one, 7(6), e38862.

Fachal L, et al. (2012) Multiple local and recent founder effects of TGM1 in Spanish families. PloS one, 7(4), e33580.

Borroni B, et al. (2011) Founder effect and estimation of the age of the Progranulin Thr272fs mutation in 14 Italian pedigrees with frontotemporal lobar degeneration. Neurobiology of aging, 32(3), 555.e1.

Semmler A, et al. (2011) Haplotype analysis of the 5,10-methylenetetrahydrofolate reductase (MTHFR) c.1298A>C (E429A) polymorphism. BMC research notes, 4, 439.

Cornes BK, et al. (2010) Haplotype analysis reveals a possible founder effect of RET mutation R114H for Hirschsprung's disease in the Chinese population. PloS one, 5(6), e10918.

Cichon S, et al. (2006) Increased activity of coagulation factor XII (Hageman factor) causes hereditary angioedema type III. American journal of human genetics, 79(6), 1098.

Patin E, et al. (2006) Deciphering the ancient and complex evolutionary history of human arylamine N-acetyltransferase genes. American journal of human genetics, 78(3), 423.

Molitor J, et al. (2004) A survey of current Bayesian gene mapping methods. Human genomics, 1(5), 371.