

Casari

Becchetti A, De Fusco M, Crociani O, Cherubini A, Restano-Cassulini R, Lecchi M, Masi A, Arcangeli A, Casari G, Wanke E.

The functional properties of the human ether-a-go-go-like (HELK2) K⁺ channel. *Eur. J. Neurosci.* 16: 415-428 (2002).

Filla A, De Michele G, Coccozza S, Patrignani A, Volpe G, Castaldo I, Ruggiero G, Bonavita V, Masters C, Casari G, Bruni A.

Early onset autosomal dominant dementia with ataxia, extrapyramidal features, and epilepsy.

Neurology 58: 922-928 (2002).

Guerrini R, Parmeggiani L, Casari G.

Epilepsy and paroxysmal dyskinesia: co-occurrence and differential diagnosis.

Adv. Neurol. 89: 433-441 (2002).

Lamantea E, Tiranti V, Bordoni A, Toscano A, Bono F, Servidei S, Papadimitriou A, Spelbrink H, Silvestri L, Casari G, Comi GP, Zeviani M.

Mutations of mitochondrial DNA polymerase gammaA are a frequent cause of autosomal dominant or recessive progressive external ophthalmoplegia.

Ann. Neurol. 52: 211-219 (2002).

Muglia M, Magariello A, Nicoletti G, Patitucci A, Gabriele AL, Conforti FL, Mazzei R, Caracciolo M, Casari G, Ardito B, Lastilla M, Gambardella A, Quattrone A.

A large family with pure autosomal dominant hereditary spastic paraplegia from southern Italy mapping to chromosome 14q11.2-q24.3.

J. Neurol. 249: 1413-1416 (2002).

Santoro L, Manganelli F, Di Maio L, Barbieri F, Carella M, D'Adamo P, Casari G.

Charcot-Marie-Tooth disease type 2C: a distinct genetic entity. Clinical and molecular characterization of the first European family.

Neuromusc. Disord. 12: 399-404 (2002).

Syren ML, Tedeschi S, Cesareo L, Bellantuono R, Colussi G, Procaccio M, Ali A, Domenici R, Malberti F, Sprocati M, Sacco M, Miglietti N, Edefonti A, Sereni F, Casari G, Coviello DA, Bettinelli A.

Identification of fifteen novel mutations in the SLC12A3 gene encoding the Na-Cl Co-transporter in Italian patients with Gitelman syndrome.

Hum. Mutat. 20: 78 (2002).

Toto L, Parodi MB, Baralle F, Casari G, Ravalico G, Romano M.

Genetic heterogeneity in Malattia Leventinese.

Clin. Genet. 62: 399-403 (2002).

Annunziata I, Lanzara C, Conte I, Zullo A, Ventruto V, Rinaldi MM, D'Urso M, Casari G, Ciccodicola A, Miano MG.

Mapping of MRX81 in Xp11.2-Xq12 suggests the presence of a new gene involved in nonspecific X-linked mental retardation.

Am. J. Med. Genet. A 118: 217-222 (2003).

Atorino L, Silvestri L, Koppen M, Cassina L, Ballabio A, Marconi R, Langer T, Casari G.

Loss of m-AAA protease in mitochondria causes complex I deficiency and increased sensitivity to oxidative stress in hereditary spastic paraplegia.

J. Cell Biol. 163: 777-787 (2003).

Bonati MT, Ferini-Strambi L, Aridon P, Oldani A, Zucconi M, Casari G.

Autosomal dominant restless legs syndrome maps on chromosome 14q.

Brain 126: 1485-1492 (2003).

Casari G, Amoroso A.

Molecular analysis of uromodulin and SAH genes, positional candidates for autosomal dominant medullary cystic kidney disease linked to 16p12.

J. Nephrol. 16: 459 (2003).

De Fusco M, Marconi R, Silvestri L, Atorino L, Rampoldi L, Morgante L, Ballabio A, Aridon P, Casari G.

Haploinsufficiency of ATP1A2 encoding the Na⁺/K⁺ pump alpha2 subunit associated with familial hemiplegic migraine type 2.

Nature Genet. 33: 192-196 (2003).

De Michele G, Maltecca F, Carella M, Volpe G, Orio M, De Falco A, Gombia S, Servadio A, Casari G, Filla A, Bruni A.

Dementia, ataxia, extrapyramidal features, and epilepsy: phenotype spectrum in two Italian families with spinocerebellar ataxia type 17.

Neurol. Sci. 24:1 66-167 (2003).

Guerrini R, Casari G, Marini C.

The genetic and molecular basis of epilepsy.

Trends Mol. Med. 9: 300-306 (2003).

Maltecca F, Filla A, Castaldo I, Coppola G, Fragassi NA, Carella M, Bruni A, Coccozza S, Casari G, Servadio A, De Michele G.

Intergenerational instability and marked anticipation in SCA-17.

Neurology 61: 1441-1443 (2003).

Marconi R, De Fusco M, Aridon P, Plewnia K, Rossi M, Carapelli S, Ballabio A, Morgante L, Musolino R, Epifanio A, Micieli G, De Michele G, Casari G.

Familial hemiplegic migraine type 2 is linked to 0.9Mb region on chromosome 1q23.

Ann. Neurol. 53 :376-381 (2003).

Monti LD, Barlassina C, Citterio L, Galluccio E, Berzuini C, Setola E, Valsecchi G, Lucotti P, Pozza G, Bernardinelli L, Casari G, Piatti P.

Endothelial nitric oxide synthase polymorphisms are associated with type 2 diabetes and the insulin resistance syndrome.

Diabetes 52: 1270-1275 (2003).

Rampoldi L, Caridi G, Santon D, Boaretto F, Bernascone I, Lamorte G, Tardanico R, Dagnino M, Colussi G, Scolari F, Ghiggeri GM, Amoroso A, Casari G.

Allelism of MCKD, FJHN and GCKD caused by impairment of uromodulin export dynamics.
Hum. Mol. Genet. 12: 3369-3384 (2003).

Scolari F, Viola BF, Ghiggeri GM, Caridi G, Amoroso A, Rampoldi L, Casari G.
Towards the identification of (a) gene(s) for autosomal dominant medullary cystic kidney disease.
J. Nephrol. 16: 321-328 (2003).

Zerbini G, Maestroni A, Breviario D, Mangili R, Casari G.
Alternative splicing of NHE-1 mediates Na-Li countertransport and associates with activity rate.
Diabetes 52: 1511-1518 (2003).

Aridon P, D'Andrea G, Rigamonti A, Leone M, Casari G, Bussone G.
Elusive amines and cluster headache: mutational analysis of trace amine receptor cluster on chromosome 6q23.
Neurol. Sci. 25 Suppl 3: S279-280 (2004).

Bassi MT, Bresolin N, Tonelli A, Nazos K, Crippa F, Baschirotto C, Zucca C, Bersano A, Dolcetta D, Boneschi FM, Barone V, Casari G.
A novel mutation in the ATP1A2 gene causes alternating hemiplegia of childhood.
J. Med. Genet. 41: 621-628 (2004).

Bruni AC, Takahashi-Fujigasaki J, Maltecca F, Foncin JF, Servadio A, Casari G, D'Adamo P, Maletta R, Curcio SA, De Michele G, Filla A, El Hachimi KH, Duyckaerts C.
Behavioral disorder, dementia, ataxia, and rigidity in a large family with TATA box-binding protein mutation.
Arch. Neurol. 61: 1314-1320 (2004).

Ferini-Strambi L, Bonati MT, Oldani A, Aridon P, Zucconi M, Casari G.
Genetics in restless legs syndrome.
Sleep Med. 5: 301-304 (2004).

Ferreirinha F, Quattrini A, Pirozzi M, Valsecchi V, Dina G, Broccoli V, Auricchio A, Piemonte F, Tozzi G, Gaeta L, Casari G, Ballabio A, Rugarli EI.
Axonal degeneration in paraplegin-deficient mice is associated with abnormal mitochondria and impairment of axonal transport.
J. Clin. Invest. 113: 231-242 (2004).

Gambara G, Vezzoli G, Casari G, Rampoldi L, D'Angelo A, Borghi L.
Genetics of hypercalciuria and calcium nephrolithiasis: from the rare monogenic to the common polygenic forms.
Am. J. Kidney Dis. 44: 963-986 (2004).

Lavorgna G, Dahary D, Lehner B, Sorek R, Sanderson CM, Casari G.
In search of antisense.
Trends Biochem. Sci. 29: 88-94 (2004).

Lavorgna G, Sessa L, Guffanti A, Lassandro L, Casari G.
AntiHunter: searching BLAST output for EST antisense transcripts.
Bioinformatics 20: 583-585 (2004).

Scolari F, Caridi G, Rampoldi L, Tardanico R, Izzi C, Pirulli D, Amoroso A, Casari G, Ghiggeri GM.
Uromodulin storage diseases: clinical aspects and mechanisms.
Am. J. Kidney Dis. 44: 987-999 (2004).

Striano P, Chifari R, Striano S, de Fusco M, Elia M, Guerrini R, Casari G, Canevini MP.
A new benign adult familial myoclonic epilepsy (BAFME) pedigree suggesting linkage to chromosome 2p11.1-q12.2.
Epilepsia 45: 190-192 (2004).

Lavorgna G, Triunfo R, Santoni F, Orfanelli U, Noci S, Bulfone A, Zanetti G, Casari G.
AntiHunter 2.0: increased speed and sensitivity in searching BLAST output for EST antisense transcripts.
Nucleic Acids Res. 33: W665-668 (2005).

Martinelli Boneschi F, Aridon P, Zara F, Guerrini R, Marini C, De Fusco M, Comi G, Casari G.
No evidence of ATP1A2 involvement in 12 multiplex Italian families with benign familial infantile seizures.
Neurosci. Lett. 388: 71-74 (2005).

Riant F, De Fusco M, Aridon P, Ducros A, Ploton C, Marchelli F, Maciazek J, Bousser MG, Casari G, Tournier-Lasserre E.
ATP1A2 mutations in 11 families with familial hemiplegic migraine.
Hum. Mutat. 26: 281 (2005).

Silvestri L, Caputo V, Bellacchio E, Atorino L, Dallapiccola B, Valente EM, Casari G.
Mitochondrial import and enzymatic activity of PINK1 mutants associated to recessive parkinsonism.
Hum. Mol. Genet. 14: 3477-3492 (2005).

Aridon P, Marini C, Di Resta C, Brilli E, De Fusco M, Politi F, Parrini E, Manfredi I, Pisano T, Pruna D, Curia G, Cianchetti C, Pasqualetti M, Becchetti A, Guerrini R, Casari G.
Increased sensitivity of the neuronal nicotinic receptor alpha 2 subunit causes familial epilepsy with nocturnal wandering and ictal fear.
Am. J. Hum. Genet. 79: 342-350 (2006).

Bernascone I, Vavassori S, Di Pentima A, Santambrogio S, Lamorte G, Amoroso A, Scolari F, Ghiggeri GM, Casari G, Polishchuk R, Rampoldi L.
Defective intracellular trafficking of uromodulin mutant isoforms.
Traffic 7: 1567-1579 (2006).

Pichler I, Marroni F, Volpato CB, Gusella JF, Klein C, Casari G, De Grandi A, Pramstaller PP.

Linkage analysis identifies a novel locus for restless legs syndrome on chromosome 2q in a South Tyrolean population isolate.

Am. J. Hum. Genet. 79: 716-723 (2006).

Vanmolkot KR, Kors EE, Turk U, Turkdogan D, Keyser A, Broos LA, Kia SK, van den Heuvel JJ, Black DF, Haan J, Frants RR, Barone V, Ferrari MD, Casari G, Koenderink JB, van den Maagdenberg AM.

Two de novo mutations in the Na,K-ATPase gene ATP1A2 associated with pure familial hemiplegic migraine.

Eur. J. Hum. Genet. 14: 555-560 (2006).

Vogl FD, Pichler I, Adel S, Pinggera GK, Bracco S, De Grandi A, Volpato CB, Aridon P, Mayer T, Meitinger T, Klein C, Casari G, Pramstaller PP.

Restless legs syndrome: epidemiological and clinicogenetic study in a South Tyrolean population isolate.

Mov. Disord. 21: 1189-1195 (2006).

Aridon P, Ragonese P, De Fusco M, Lo Coco D, Salemi G, Casari G, Savettieri G.

Autosomal dominant hereditary spastic paraplegia: report of a large Italian family with R581X spastin mutation.

Neurol. Sci. 28: 171-174 (2007).

Colussi G, Bettinelli A, Tedeschi S, De Ferrari ME, Syren ML, Borsa N, Mattiello C, Casari G, Bianchetti MG.

A thiazide test for the diagnosis of renal tubular hypokalemic disorders.

Clin. J. Am. Soc. Nephrol. 2: 454-460 (2007).

Del Chiaro M, Zerbi A, Falconi M, Bertacca L, Polese M, Sartori N, Boggi U, Casari G, Longoni BM, Salvia R, Caligo MA, Di Carlo V, Pederzoli P, Presciuttini S, Mosca F.

Cancer risk among the relatives of patients with pancreatic ductal adenocarcinoma.

Pancreatology 7: 459-469 (2007).

Koppen M, Metodiev MD, Casari G, Rugarli EI, Langer T.

Variable and tissue-specific subunit composition of mitochondrial m-AAA protease complexes linked to hereditary spastic paraplegia.

Mol. Cell. Biol. 27:758-767 (2007).

Sessa L, Breiling A, Lavorgna G, Silvestri L, Casari G, Orlando V.

Noncoding RNA synthesis and loss of Polycomb group repression accompanies the colinear activation of the human HOXA cluster.

RNA 13: 223-239 (2007).

Aridon P, Ragonese P, De Fusco M, Salemi G, Casari G, Savettieri G.

Further evidence of genetic heterogeneity in familial essential tremor.

Parkinsonism Relat. Disord. 14: 15-18 (2008).

Galluccio E, Piatti P, Citterio L, Lucotti PC, Setola E, Cassina L, Oldani M, Zavaroni I, Bosi E, Colombo A, Alfieri O, Casari G, Reaven GM, Monti LD.
Hyperinsulinemia and impaired leptin-adiponectin ratio associate with endothelial nitric oxide synthase polymorphisms in subjects with in-stent restenosis.
Am. J. Physiol. 294: E978-986 (2008).

Hansen JM, Thomsen LL, Marconi R, Casari G, Olesen J, Ashina M.
Familial hemiplegic migraine type 2 does not share hypersensitivity to nitric oxide with common types of migraine.
Cephalalgia 28: 367-375 (2008).

Madia F, Striano P, Di Bonaventura C, de Falco A, de Falco FA, Manfredi M, Casari G, Striano S, Minetti C, Zara F.
Benign adult familial myoclonic epilepsy (BAFME): evidence of an extended founder haplotype on chromosome 2p11.1-q12.2 in five Italian families.
Neurogenetics 9: 139-142 (2008).

Maltecca F, Aghaie A, Schroeder DG, Cassina L, Taylor BA, Phillips SJ, Malaguti M, Previtali S, Guenet JL, Quattrini A, Cox GA, Casari G.
The mitochondrial protease AFG3L2 is essential for axonal development.
J. Neurosci. 28: 2827-2836 (2008).

De Grandi A, Volpato CB, Bedin E, Pattaro C, Marroni F, Pichler I, Hicks AA, Casari G, Pramstaller PP.
ParkScreen: A Low-Cost Rapid Linkage Marker Panel for Parkinson's Disease.
J. Mol. Neurosci. (2009).

Kathiresan S, Voight BF, Purcell S, Musunuru K, Ardissino D, Mannucci PM, Anand S, Engert JC, Samani NJ, Schunkert H, Erdmann J, Reilly MP, Rader DJ, Morgan T, Spertus JA, Stoll M, Girelli D, McKeown PP, Patterson CC, Siscovick DS, O'Donnell CJ, Elosua R, Peltonen L, Salomaa V, Schwartz SM, Melander O, Altshuler D, Ardissino D, Merlini PA, Berzuini C, Bernardinelli L, Peyvandi F, Tubaro M, Celli P, Ferrario M, Fritzsche J, Marziliano N, Casari G, et al.
Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants.
Nature Genet. 41: 334-341 (2009).

Manfredi I, Zani AD, Rampoldi L, Pegorini S, Bernascone I, Moretti M, Gotti C, Croci L, Consalez GG, Ferini-Strambi L, Sala M, Pattini L, Casari G.
Expression of mutant beta2 nicotinic receptors during development is crucial for epileptogenesis.
Hum. Mol. Genet. 18: 1075-1088 (2009).

Martinelli P, La Mattina V, Bernacchia A, Magnoni R, Cerri F, Cox G, Quattrini A, Casari G, Rugarli EI.
Genetic interaction between the m-AAA protease isoenzymes reveals novel roles in cerebellar degeneration.
Hum. Mol. Genet. 18: 2001-2013 (2009).

Schaeffer C, Santambrogio S, Perucca S, Casari G, Rampoldi L.

Analysis of uromodulin polymerization provides new insights into the mechanisms regulating ZP domain-mediated protein assembly.
Mol. Biol. Cell 20: 589-599 (2009).

Maltecca F, Magnoni R, Cerri F, Cox GA, Quattrini A, Casari G.
Haploinsufficiency of AFG3L2, the gene responsible for spinocerebellar ataxia type 28 (SCA 28), causes mitochondria-mediated Purkinje cell dark degeneration.
J.Neurosci. in press (2009).