ARTICLES

Partial complex I deficiency due to the CNS conditional ablation of Ndufa5 results in a mild chronic encephalopathy but no increase in oxidative damage

S. Peralta, A. Torraco, T. Wenz, S. Garcia, F. Diaz, and C.T. Moraes

Abnormal mitochondrial transport and morphology are common pathological denominators in SOD1 and TDP43 ALS mouse models

J. Magrané, C. Cortez, W.-B. Gan, and G. Manfredi

The CYP27B1 variant associated with an increased risk of autoimmune disease is underexpressed in tolerizing dendritic cells

F. Shahijanian, G.P. Parnell, F.C. McKay, P.N. Gatt, M. Shojoie, K.S. O’Connor, S.D. Schibeci, F. Brilot, C. Liddle, M. Batten, ANZgene Multiple Sclerosis Genetics Consortium, G.J. Stewart, and D.R. Booth

FOXO3 determines the accumulation of α-synuclein and controls the fate of dopaminergic neurons in the substantia nigra

E. Pino, R. Amamoto, L. Zheng, M. Cacquevel, J.-C. Sarria, G.W. Knott, and B.L. Schneider

Different mtDNA mutations modify tumor progression in dependence of the degree of respiratory complex I impairment

L. Iommarini, I. Kurelac, M. Capristo, M.A. Calvaruso, V. Giorgio, C. Bergamini, A. Ghelli, P. Nanni, C. De Giovanni, V. Carelli, R. Fato, P.L. Lollini, M. Rugolo, G. Gasparre, and A.M. Porcelli

Targeted manipulation of the sortilin–progranulin axis rescues progranulin haploinsufficiency

W.C. Lee, S. Almeida, M. Prudencio, T.R. Caulfield, Y.-J. Zhang, W.M. Tay, P.O. Bauer, J. Chew, H. Sasaguri, K.R. Jansen-West, T.F. Gendron, C.T. Stetler, N.C. Finch, I.R. Mackenzie, R. Rademakers, F.-B. Gao, and L. Petrucelli

Absence of cell surface expression of human ACE leads to perinatal death

A. Michaud, K.R. Acharya, G. Masuyer, N. Quenech’du, O. Gribouval, V. Morinère, M.-C. Gubler, and P. Corvol

Distinct roles of TRAF6 at early and late stages of muscle pathology in the mdx model of Duchenne muscular dystrophy

S.M. Hindi, S. Sato, Y. Choi, and A. Kumar

New Lmna knock-in mice provide a molecular mechanism for the ‘segmental aging’ in Hutchinson–Gilford progeria syndrome

H.-J. Jung, Y. Tu, S.H. Yang, A. Tatar, C. Nobumori, D. Wu, S.G. Young, and L.G. Fong

Beta tubulin isoforms are not interchangeable for rescuing impaired radial migration due to Tubb3 knockdown

Y. Saillour, L. Broix, E. Bruel-Jungerman, N. Lebrun, G. Muraca, J. Rucci, K. Poirier, R. Belvindrah, F. Francis, and J. Chelly

Inferring primary tumor sites from mutation spectra: a meta-analysis of histology-specific aberrations in cancer-derived cell lines

F. Dietlein and W. Eschner
Mosaic synaptopathy and functional defects in Cav1.4 heterozygous mice and human carriers of CSNB2

S. Michalakis, L. Shaltiel, V. Sothilingam, S. Koch, V. Schludi, S. Krause, C. Zeitz, I. Audo, M.-E. Lancelot, C. Hamel, I. Meunier, M.N. Preising, C. Friedburg, B. Lorenz, N. Zabouri, S. Haverkamp, M.G. Garrido, N. Tanimoto, M.W. Seeliger, M. Biel, and C.A. Wahl-Schott

High-content screening identifies small molecules that remove nuclear foci, affect MBNL distribution and CELF1 protein levels via a PKC-independent pathway in myotonic dystrophy cell lines

A. Ketley, C.Z. Chen, X. Li, S. Arya, T.E. Robinson, J. Granados-Riveron, I. Udosen, G.E. Morris, I. Holt, D. Furling, S. Chaouch, B. Haworth, N. Southall, P. Shinn, W. Zheng, C.P. Austin, C.J. Hayes, and J.D. Brook

Epigenetic dysregulation of SHANK3 in brain tissues from individuals with autism spectrum disorders

L. Zhu, X. Wang, X.-L. Li, A. Towers, X. Cao, P. Wang, R. Bowman, H. Yang, J. Goldstein, Y.-J. Li, and Y.-H. Jiang

Major epigenetic development distinguishing neuronal and non-neuronal cells occurs postnatally in the murine hypothalamus

G. Li, W. Zhang, M.S. Baker, E. Laritsky, N. Mattan-Hung, D. Yu, G. Kunde-Ramamoorthy, R.B. Simerly, R. Chen, L. Shen, and R.A. Waterland

Methionine sulfoxide reductase B3 deficiency causes hearing loss due to stereocilia degeneration and apoptotic cell death in cochlear hair cells

T.-J. Kwon, H.-J. Cho, U.-K. Kim, E. Lee, S.-K. Oh, J. Bok, Y.C. Bae, J.-K. Yi, J.W. Lee, Z.-Y. Ryoo, S.H. Lee, K.-Y. Lee, and H.-Y. Kim

A new congenital disorder of glycosylation caused by a mutation in SSR4, the signal sequence receptor 4 protein of the TRAP complex

M.E. Losfield, B.G. Ng, M. Kircher, K.J. Buckingham, E.H. Turner, A. Eroshkin, J.D. Smith, J. Shendure, D.A. Nickerson, M.J. Bamshad, University of Washington Center for Mendelian Genomics, and H.H. Freeze

Expression levels of DNA replication and repair genes predict regional somatic repeat instability in the brain but are not altered by polyglutamine disease protein expression or age

A.G. Mason, S. Tomé, J.P. Simard, R.T. Libby, T.K. Bammler, R.P. Beyer, A.J. Morton, C.E. Pearson, and A.R. La Spada

SHOX triggers the lysosomal pathway of apoptosis via oxidative stress

G. Hristov, T. Marttila, C. Durand, B. Niesler, G.A. Rappold, and A. Marchini

Dual-tagged amyloid-β precursor protein reveals distinct transport pathways of its N- and C-terminal fragments

C. Villegas, V. Muresan, and Z. Ladescu Muresan

SIRT2 regulates ciliogenesis and contributes to abnormal centrosome amplification caused by loss of polycystin-1

X. Zhou, L.X. Fan, K. Li, R. Ramchandran, J.P. Calvet, and X. Li

ASSOCIATION STUDIES ARTICLES

Allelic heterogeneity in NCF2 associated with systemic lupus erythematosus (SLE) susceptibility across four ethnic populations

X. Kim-Howard, C. Sun, J.E. Molineros, A.K. Maiti, H. Chandru, A. Adler, G.B. Wiley, K.M. Kaufman, L. Kottyan, J.M. Guthridge, A. Rasmussen, J. Kelly, E. Sánchez, P. Raj, Q.-Z. Li, S.-Y. Bang, H.-S. Lee, T.-H. Kim, Y.M. Kang, C.-H. Suh, W.T. Chung, Y.-B. Park, J.-Y. Choe, S.C. Shim, S.-S. Lee, B.-G. Han, N.J. Olsen, D.R. Karp, K. Moser, B.A. Pons-Estel, E.K. Wakeland, J.A. James, J.B. Harley, S.-C. Bae, P.M. Gaffney, M. Alarcón-Riquelme, on behalf of GENLES, L.L. Looger, and S.K. Nath
CNV analysis in a large schizophrenia sample implicates deletions at 16p12.1 and SLC1A1 and duplications at 1p36.33 and CGNL1

E. Rees, J.T.R. Walters, K.D. Chambert, C. O’Dushlaine, J. Szatkiewicz, A.L. Richards, L. Georgieva, G. Mahoney-Davies, S.E. Legge, J.L. Moran, G. Genovese, D. Levinson, D.W. Morris, P. Cormican, K.S. Kendler, F.A. O’Neill, B. Riley, M. Gill, A. Corvin, Wellcome Trust Case Control Consortium, P. Sklar, C. Hultman, C. Pato, M. Pato, P.F. Sullivan, P.V. Gejman, S.A. McCarroll, M.C. O’Donovan, M.J. Owen, and G. Kirov

Cover: To determine the basis of trinucleotide repeat instability differences between brain regions, Mason et al. surveyed the transcriptome of the striatum (high levels of instability) and the cerebellum (low levels of instability) in adult C57BL/6J mice. The left panel presents a heat map of microarray expression correlations for six cerebellar samples (C1–C6) and six striatal samples (S1–S6). Strong correlations occur between samples from the same tissue, while correlations are weak between different tissues. To delineate protein interactions centered at related genes with the greatest expression differences, STRING analysis was performed and yielded the interaction network shown in the right panel. See article by A. G. Mason et al., 1606–1618.