ARTICLES

A non-human primate system for large-scale genetic studies of complex traits

Enhanced J-protein interaction and compromised protein stability of mtHsp70 variants lead to mitochondrial dysfunction in Parkinson’s disease
A.V. Goswami, M. Samaddar, D. Sinha, J. Purushotham, and P. D’Silva

OCRL localizes to the primary cilium: a new role for cilia in Lowe syndrome

Small rare recurrent deletions and reciprocal duplications in 2q21.1, including brain-specific ARHGEF4 and GPR148

Reticulon-like-1, the Drosophila orthologue of the Hereditary Spastic Paraplegia gene reticulon 2, is required for organization of endoplasmic reticulum and of distal motor axons

Accumulation of instability in serial differentiation and reprogramming of parthenogenetic human cells
R. Vassena, N. Montserrat, B. Carrasco Canal, B. Aran, L. de Onate, A. Veiga, and J.C. Izpisua Belmonte

A t(1;11) translocation linked to schizophrenia and affective disorders gives rise to aberrant chimeric DISC1 transcripts that encode structurally altered, deleterious mitochondrial proteins

MET expression in melanoma correlates with a lymphangiogenic phenotype
A. Swoboda, O. Schanab, S. Tauber, M. Bilban, W. Berger, P. Petzelbauer, and M. Mikula

Loss of GM3 synthase gene, but not sphingosine kinase 1, is protective against murine nephronophthisis-related polycystic kidney disease

Disease severity in a mouse model of ataxia telangiectasia is modulated by the DNA damage checkpoint gene Hus1
Mutant superoxide dismutase 1 (SOD1), a cause of amyotrophic lateral sclerosis, disrupts the recruitment of SMN, the spinal muscular atrophy protein to nuclear Cajal bodies

Molecular mechanisms of riboflavin responsiveness in patients with ETF-QO variations and multiple acyl-CoA dehydrogenation deficiency

Variable phenotype of del45-55 Becker patients correlated with nNOS\(_\alpha\) mislocalization and RYR1 hypernitrosylation

A greatly extended PPARGC1A genomic locus encodes several new brain-specific isoforms and influences Huntington disease age of onset

Transducer of regulated CREB-binding proteins (TORCs) transcription and function is impaired in Huntington’s disease

Alteringons of social interaction through genetic and environmental manipulation of the 22q11.2 gene Sept5 in the mouse brain

ASSOCIATION STUDIES ARTICLES

Evidence for a role of the rare p.A152T variant in MAPT in increasing the risk for FTD-spectrum and Alzheimer’s diseases

Evaluation of copy number variations reveals novel candidate genes in autism spectrum disorder-associated pathways

Cover: Alcian blue- and Alizarin red-stained skulls from Hus1\(^{+/neoAm}^{+/}\) (top) and Hus1\(^{nea/neo}Am^{--/}\) (bottom) mouse embryos at E18.5. In these preparations, cartilage is stained blue and bone is stained red. The skull from the Hus1\(^{nea/neo}Am^{--/}\) embryo is smaller and presents an array of fenestrations in the parietal bone, highlighting the craniofacial defects present in Atm/Hus1 double mutant mice. For further detail, see the article by Balmus et al., pp. 3408–3420.