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physiology to the clinical features of patients with neurological disease. . Professor Pamela J Shaw MBBS MD FRCP FAAN FANA FMedSci of the Sheffield Care and Research Centre for Motor Neuron Disorders; . models, patient material and neuroimaging to understand the molecular, cellular and genetic aspects.

Chromosome 2q37 deletion: clinical and molecular

How to Cite. Falk, R. E. and Casas, K. A. (2007), Chromosome 2q37 deletion: Clinical and molecular aspects. Am. J. Med. Genet., 145C: 357-371. doi: 10.1002/ajmg.c.30153

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Kokotas H, Petersen MB. Clinical and molecular aspects of aniridia. Clin Genet. 2010 May;77(5):409-20.

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induced hyperammonemia without clinical hepatic dysfunction. Neurology 1982 Pamela J. Shaw, MD, FRCP, ". Paul G. Ince, MD, patients with sporadic motor neuron disease, 8 neurolog- tions, the molecular mechanisms of motor neuron in- jury and .. vival, or unique aspects of glutamatergic neurotrans- mission in

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Jan 20, 2012 Johnathan Cooper-Knock, Pamela J. Shaw and Janine Kirby. Sheffield . mirror aspects of the disease process and enable mechanistic insights and therapeutic approaches to be . motor neurone involvement and a slow clinical progression. The gene .. Human molecular genetics 15:233-50. Hand CK

The widening spectrum of c9orf72 related disease

Feb 4, 2014 phenotype. Johnathan Cooper Knock Pamela J. Shaw . Janine Kirby ferling clinical phenotypes arising from the mutation. Keywords .. neurone disease (FTLD ALS/FTLD MND), which will be discussed later. . The molecular basis for this disease .. ular Aspects of Motor Neuron Disease. Colloq Ser

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Correspondence to: Professor Pamela J Shaw Academic Neurology Unit, The pathology accompanying the clinical phenotype was first described by Jean- Martin Charcot in 1869. Molecular mechanisms that may contribute to motor neurone injury in motor neurone disease. Clinical aspects of sporadic ALS/ MND.

Sheffield experimental medicine programmes in neuroscience

Professor Pamela Shaw, Professor of Neurology University of Sheffield, Honorary application of computation modelling to all aspects of clinical neuroscience research. in survival and quality of life in patients with motor neurone disease (MND). elucidating the molecular pathophysiology of these diseases [46-50].

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Angelman syndrome: review of clinical and molecular aspects Lynne M Bird¹Department of Pediatrics, University of California, Division of Genetics, Rady Children s

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Mar 22, 2008 Christopher J McDermott, clinical senior lecturer and honorary consultant neurologist and Pamela J Shaw, professor We will discuss the presentation of motor neurone disease in primary care and update .. for patients, carers, and UK healthcare professionals on all aspects of motor neurone disease.

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How to Cite. Kokotas, H. and Petersen, M. (2010), Clinical and molecular aspects of aniridia. *Clinical Genetics*, 77: 409-420. doi: 10.1111/j.1399-0004.2010.01372.x

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219 disease (PiD) and the intraneuronal inclusions became

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Mar 3, 2015 Johnathan Cooper-Knock, Janine Kirby, Robin Highley, and Pamela J. Shaw corresponding author. Sheffield. Clinical Phenotypes Associated with the C9orf72 Expansion in ALS Cooper-Knock J, Shaw PJ, Kirby J. The widening spectrum of Clinical and molecular aspects of motor neuron disease.

Disaccharide digestion: clinical and molecular

1. *Clin Gastroenterol Hepatol*. 2006 Mar;4(3):276-87. Disaccharide digestion: clinical and molecular aspects. Robayo-Torres CC(1), Quezada-Calvillo R, Nichols BL.

Thyrotoxic periodic paralysis: clinical and

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Aug 25, 2014 The Knowing Neurons team accepted the ALS Ice Bucket Challenge! Ferraiuolo L., Andrew J. Grierson, Michael Sendtner & Pamela J. Shaw (2011). Molecular pathways of motor neuron injury in amyotrophic lateral sclerosis, *Clinical genetics of amyotrophic lateral sclerosis: what do we really know?*,

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Apr 17, 2015 Dame Pamela J. Shaw. A translational neuroscience approach to the problem of motor neurone disease . Clinical Affective Neuroscience . rupture in the anterior part of the circle of Willis and to explore different aspects .. Nencki Institute of Experimental Biology, Laboratory of Molecular Basis of

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