

14th International Congress of Inborn Errors of Metabolism

2021

Transforming Rare Disorders



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Pathogenic PDE12 variants impair mitochondrial RNA processing causing neonatal-onset mitochondrial disease	Dr Jose Abdenur
Case series of Wolman disease in Indonesia: recognizing diagnostic and treatment challenges	Dr Mars Nashrah Abdullah
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Clinical, pathological and molecular spectrum of patients with Glycogen Storage Diseases (GSDs) - First report from Pakistan	Dr Sibtain Ahmed
Subacute tyrosinemia type 1 cannot be detected by NBS in Japan	Dr Sayaka Ajihara
Quality of life assessment in mucopolysaccharidosis patients treated with enzyme replacement therapy	Dr Mariem Ajili
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NDUFS6 mutations cause lethal neonatal mitochondrial complex I deficiency and distinct clinical-MRI pattern	Dr Amal Alhashem
SLC25A19 deficiency: a treatable thiamine metabolic disorder	Dr Amal Alhashem
COXPD9 in an individual from Puerto Rico and literature review	Dr Hind Alsharhan
Early diagnosis of classic homocystinuria in Kuwait through newborn screening: a 6-year experience	Dr Hind Alsharhan
Gastrointestinal involvement at the junction of Wolman Disease and COVID-19	Mrs Ayşe Nur Altun
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Impact of metabolic control in phenylketonuric patients and its relationship with neurological outcomes	Prof Alvaro Ameijeiras
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Nutrition status of Brazilian patients with organic acidemias: A twenty-five year experience at a single reference center	Mr José Araújo de Oliveira Silva
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Can abetalipoproteinemia prevent the development of clinical findings of Niemann pick type C2; a case of cholesterol traffic	Dr Bengü Arslan
Kinetic analysis of N-Deacetylase/N-sulfotransferase (NDST1) inhibitors using a fluorometric coupled enzyme assay	Mr Joshua Atienza
Sapropterin testing in a paediatric centre – the Queensland experience of testing children under 18 years old for a tetrahydrobiopterin responsive form of phenylketonuria	Ms Catherine Atthow
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