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The European Society of Human Genetics

HYBRID CONFERENCE

JUNE 11–14, 2022
VIENNA, Austria

Hybrid Poster Presentation

Two de novo pathogenic variants in different genes identified by a whole exome sequencing TRIO approach explain and complement the phenotype in a patient with syndromic intellectual disability and autism.

E. Barroso[1], G. Cartagena[1], M. Martínez-Matilla[1], J.J. Ferre-Fernández[1], A. Velo[1], M. Berbel[1], R. García-Jiménez[1], I. Boyko[1], S. García-Herrero[1], C. Fernández-Vizcaino[1], D. Sánchez-Valero[1], C. Pérez-García[1], A. Fernández-Jaén[2], J. García-Planells[1].

1. GPDx Department, Igenomix S.L.
2. Department of Pediatric Neurology. Hospital Universitario Quirón Salud Madrid

Can genetic testing in healthy individuals prevent inherited cancer? Genetic data analysis of a Spanish cohort.

C. Pérez-García[1], J. Pérez-López[1], D. Sánchez-Valero[1], R. García-Jiménez[1], G. Cartagena[1], E. Barroso[1], M. Martínez-Matilla[1], S. García-Herrero[1], J. García Planells[1].

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Digital Poster Presentation

WES (Whole Exome Sequencing) diagnostic revealed the absence of SRY gene in a patient with a 47,XXY karyotype and a female phenotype.

S. García-Herrero[1], G. Cartagena[1], M. Martínez-Matilla[1], J.J. Ferre-Fernández[1], A. Velo[1], M. Berbel[1], R. García-Jiménez[1], I. Boyko[1], E. Barroso[1], C. Fernández-Vizcaino[1], D. Sánchez-Valero[1], C. Pérez-García[1], Dandalo R[1], J. García-Planells[1].

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The prevalence of X-linked inherited diseases carrier in a large Spanish cohort

J. Pérez-López[1], C. Pérez-García[1], E. Barroso[1], J. García-Planells[1].

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Early-onset atypical rare disorders: Precision genetic diagnosis aided phenotypic expansion to the rescue!

L. S Matsa, M.Sc. Ph.D.[1], SHC Appikonda, M.Sc.[1], N. John, M.Sc., Ph.D.[1], S. Elewisy, M.Sc.[1], A. El-Hattab, MD, FAAP, FACMG[1].

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The divergent pattern of inheritance in siblings with novel homozygous DMN1 gene variant

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