GASTROEXOMA

Diagnóstico etiológico de pacientes con sintomatología intestinal, hepática o pancreática.
Incluye síndromes y enfermedades del metabolismo cuyo curso clínico incluya disfunción del sistema intestinal, hepático o pancreático.

Gastroexoma analiza simultáneamente las zonas codificantes de 273 genes*, utilizando tecnologías de secuenciación masiva y protocolos bioinformáticos de diseño propio.
Nuestro análisis bioinformático exclusivo nos permite detectar variantes puntuales (SNVs), deleciones e inserciones (INDELs) y variantes en el número de copias (CNVs). Las variantes detectadas son filtradas, priorizadas y clasificadas de acuerdo con las recomendaciones científicas internacionales.

*Para un listado completo de los genes, contacte con nosotros

273 genes
~200X cobertura media
SNVs | INDELs | CNVs
Respuesta en 2-8 semanas

Subpaneles:

Páncreas
Insuficiencia pancreática
Pancreatitis
Fibrosis quística

Hígado

Colestasis
Enfermedad del hígado poliquístico
Defectos en la síntesis de ácidos biliares
Cirrosis congénita
Síndrome del fallo hepático infantil
Hemocromatosis
Enfermedad de Wilson

Intestino

Enfermedad inflamatoria intestinal (IBD)
Enfermedad de Crohn
Colitis ulcerosa

Patients with rare diseases are often disadvantaged, particularly those with rare liver diseases. Reasons for disadvantage include delayed or overlooked diagnosis, lack of local expertise and high-quality care, poor scientific understanding of the disease process and limited therapeutic options. In adult liver disease this can be compounded by prejudices towards patients with liver disease in general, because of a perception (incorrect for all rare liver diseases) that liver disease is lifestyle related and thus "self-inflicted". In paediatric rare liver diseases, such as biliary atresia, optimising outcomes requires a particularly timely diagnosis. Irrespective of patient age, the scientific and medical community must rise to the challenge of advancing our understanding of rare liver disease, searching for more effective and specific therapies, and providing the infrastructure to provide the best care for all patients, infants, children, young and older adults. The European Reference Network for Rare Liver Diseases is an important step in this direction.

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David E J Jones Journal of Hepatology 2017

The prospect of genomics in IBD is that early diagnosis in a small proportion of patients might prevent unnecessary operations, severe infections, or tumors, and allow progression to targeted therapies

Holm H Uhlig Trends in Genetics 2017

genome4

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