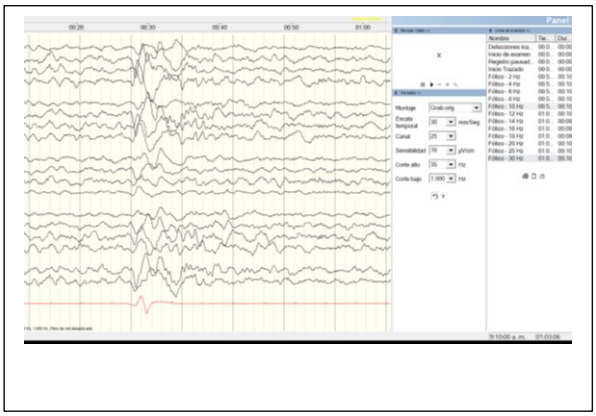
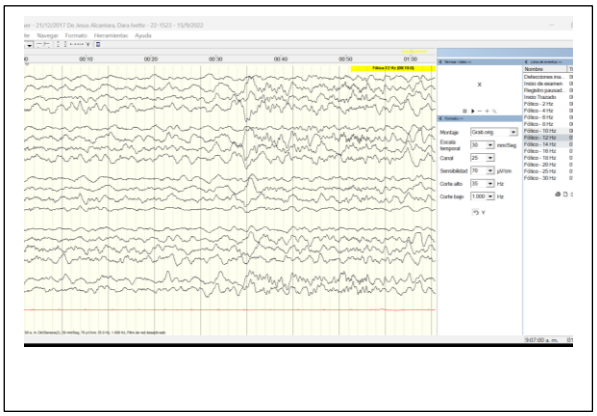
Figura suplementaria 1. Electroencefalograma.

Supplementary Figure 1. Electroencephalogram.

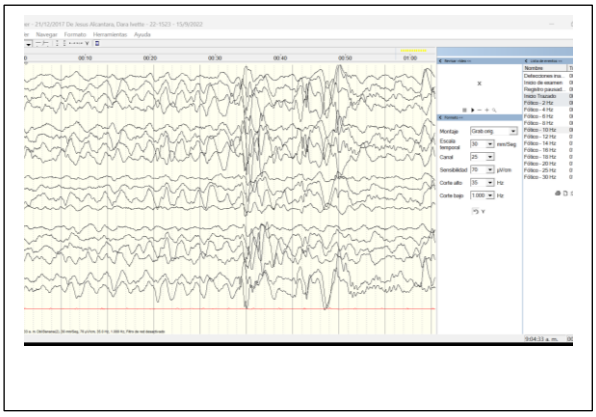
A.



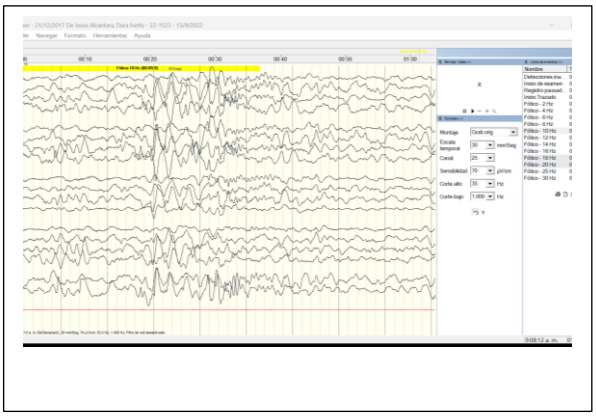
B.



C.



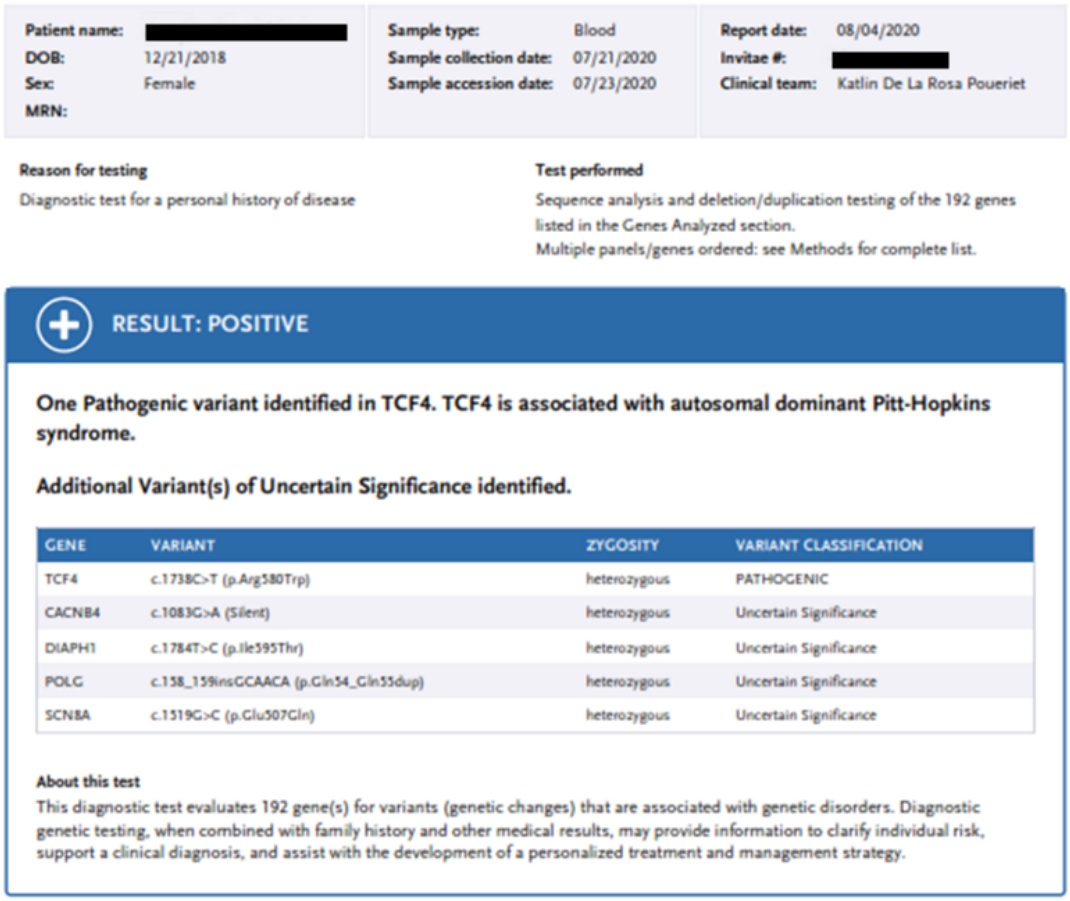
D.



Fuente/Source. Instituto ChromoMED

Figura suplementaria 2. Resultados de secuenciación.

Supplementary Figure 2. Sequencing results.



Patient name: [redacted]
DOB: 12/21/2018
Sex: Female
MRN:

Sample type: Blood
Sample collection date: 07/21/2020
Sample accession date: 07/23/2020

Report date: 08/04/2020
Invitae #: [redacted]
Clinical team: Katlin De La Rosa Poueriet

Reason for testing
Diagnostic test for a personal history of disease

Test performed
Sequence analysis and deletion/duplication testing of the 192 genes listed in the Genes Analyzed section.
Multiple panels/genes ordered: see Methods for complete list.

RESULT: POSITIVE

One Pathogenic variant identified in TCF4. TCF4 is associated with autosomal dominant Pitt-Hopkins syndrome.

Additional Variant(s) of Uncertain Significance identified.

GENE	VARIANT	ZYGOSITY	VARIANT CLASSIFICATION
TCF4	c.1738C>T (p.Arg580Trp)	heterozygous	PATHOGENIC
CACNB4	c.1083G>A (Silent)	heterozygous	Uncertain Significance
DIAPH1	c.1784T>C (p.Ile595Thr)	heterozygous	Uncertain Significance
POLG	c.158_159insGCAACA (p.Gln54_Gln55dup)	heterozygous	Uncertain Significance
SCN8A	c.1519G>C (p.Glu507Gln)	heterozygous	Uncertain Significance

About this test
This diagnostic test evaluates 192 gene(s) for variants (genetic changes) that are associated with genetic disorders. Diagnostic genetic testing, when combined with family history and other medical results, may provide information to clarify individual risk, support a clinical diagnosis, and assist with the development of a personalized treatment and management strategy.

Figura suplementaria 3.

Supplementary Figure 3.