Technical Appendix:

Methods:

- **Libraries**: Inne panel capture (Twist)
- **Sequencing**: NextSeq500 (Illumina)
- **Bioinformatics**:
    - Alignment to the hg18 human genome reference sequence (BWA)
    - Post-alignment processing of the data according to the Broad Institute recommendations
    - Variant calling [GATK (Haplotypecaller, Unifigenotyper, Samtools and Freebayes)]
    - Search for exonic deletions and duplications (comparison of reading depths after normalization)
- **Annotation**: local interface (PolyWeb) developed by the bioinformatics platform of the University of Paris and Imagine
- **Databases used**: gnomAD, HGMD-Pro, Clinvar
- **Prediction software**: Polyphen-2 HumanVar (PPN2), SIFT, Mutation Taster, CAD
- Current HGVS nomenclature

Comments and limitations:

Gene coverage is defined as coding and flanking intronic sequences (+/- 20 bp)
Exons with no or poor coverage (< 3°X) are not sequenced by any other method.
Variants classified as benign or probably benign are not shown in this report.
Single heterozygous variants identified in genes involved in autosomal recessive pathology are not reported.
A negative result does not exclude a mutation not detected by the technique used.

List of poorly covered exons:

TBX1: exon 3

List of genes in the Inne panel:

BCL2G (NM003922)
CARD9 (NM_052813)
CARML2 (NM_001013838)
CD40LG (NM_000074)
CYBA (NM_000101)
CYBB (NM_000397)
CYBC1 (NM_001078087)
DOCKB (NM_203447)
GATA2 (NM_032638)
INFg (NM_000619)
INFGR1 (NM_000416)
INFGR2 (NM_001329128)
IKBKB (NM_001556)

IKBKG (NM_001099856)
IL129 (NM_002187)

IL12RBI (NM_005535)
IL12RB2 (NM_001559)
IL17A (NM_002190)
IL17F (NM_052872)
IL17RA (NM_014339)
IL17RC (NM_153461)
IL23R (NM_142701)
IRAK4 (NM_016123)
IRF1 (NM_002198)
IRF4 (NM_002460)
IRF8 (NM_002153)

ISG15 (NM_005101)
JAK1 (NM_002227)
MAPKB (NM_139049)
MYD88 (NM_001172567)
NCF1 (NM_000265)
NCF2 (NM_000433)
NCF4 (NM_013416)
NFKB1 (NM_001077 34)
NFKB1A (NM_020529)
PRKCD (NM_006254)
REL (NM_002908)
RORC (NM_005060)

SLC11A1 (NM_000578)
SPPL2A (NM_032802)
STAT1 (NM_007315)
STAT2 (NM_005419)
STAT3 (NM_139276)
SYK (NM_003177)
TBX1 (NM_080647)
TBX21 (NM_013351)
TLR3 (NM_003155)
TRAF3 (NM_145725)
TRAF3NP2 (NM_147200)
TYK2 (NM_003331)
USP18 (NM_017414)