Supporting Information

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SI Methods

DNA Sample Preparation. DNA samples were obtained by direct extraction from K3 EDTA preserved whole blood (QIAamp DNA Blood Maxi Kit; Qiagen), skin fibroblast primary cultures (TRIzol Reagent; Life Technologies), or EBV-transformed lymphoblastoid lines (QIAamp DNA Mini Kit; Qiagen), generated as described (1). DNA was concentrated and measured by picogreen (Quant-iT dsDNA Assay Kit, broad range and Qubit Fluorometric Quantitation; Life Technologies). Samples showing fragmented genomic DNA by electrophoresis in 1% agarose gels were purified and concentrated (DNA Clean & Concentrator-5; Zymo Research).

Variant Validation. Selected sequence variants were validated by using Sanger sequencing on proband and parental DNA samples, where available. Variant-specific primers were designed by using the National Center for Biotechnology Information Primer-BLAST (ncbi.nlm.nih.gov/tools/primer-blast). Sequencing products were resolved on the Applied Biosystems 3730xl DNA Analyzer.

1. Anderson MA, Gusella JF (1984) Use of cyclosporin A in establishing Epstein-Barr virus-transformed human lymphoblastoid cell lines. *In Vitro* 20(11):856–858.

Other Supporting Information Files

Dataset S1 (XLSX)
Dataset S2 (XLSX)
Dataset S3 (XLSX)
Dataset S4 (XLSX)