



12,190

67 Subjects

Figure S1. Exome sequencing analysis and variant filtering scheme. Our candidate gene prioritization method included: (1) analysis of genetic polymorphisms in coding regions (exonic and splicing donor/acceptor sites) of a pre-specified list of 2,016 genes of interest (see Supplemental Table S1) that affected its protein product, including protein-truncating variants (stop gain/loss, start loss, or frameshift), missense variants, canonical splicesite variants, and inframe insertions and deletions: (2) use of Combined Annotation Dependent Depletion-based scoring, a computational protein function prediction algorithm which scores the deleteriousness of single nucleotide variants as well as INDEL variants in the human genome; and (3) selection of rare variants with a minor allele frequency <1%. Abbreviations: CADD, Combined Annotation Dependent Depletion; GOI, genes of interest; INDELs, insertions and deletions; MAF, minor allele frequency.

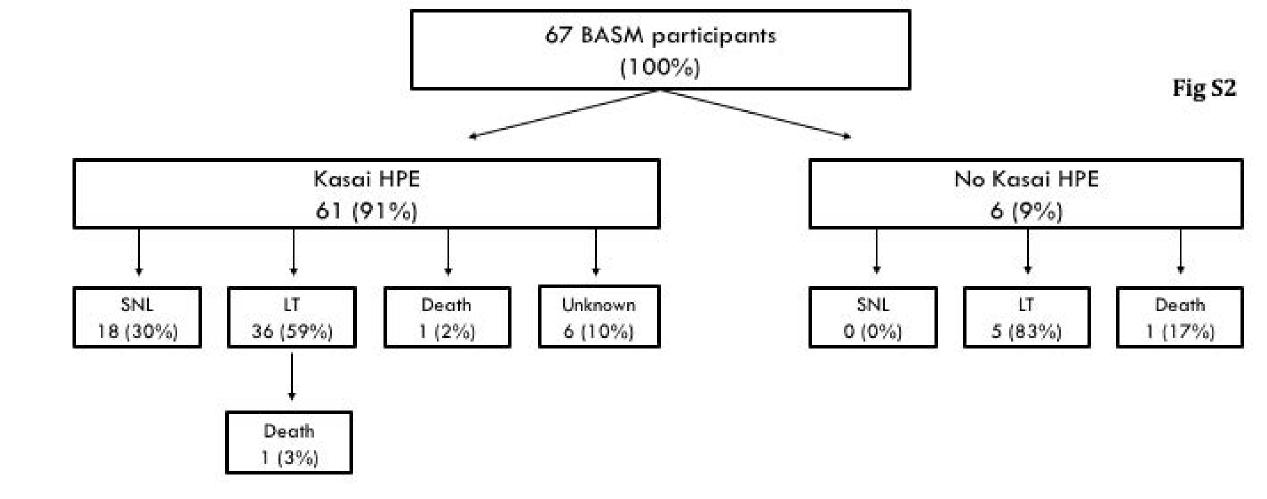


Figure S2. Flow diagram summarizing the natural history and outcomes of participants in this study at the time of selection. A designation as Unknown is defined as those without a recorded encounter beginning 12 months prior to selection date. Note that combining those with and without a Kasai HPE leads to an overall SNL rate at enrolment of 27% (18/67). Abbreviations: HPE, hepatoportoenterostomy; SNL, survival with native liver; LT, liver transplantation.