

Supplemental data

Supplemental Table S1: Provided details on whole exome sequencing and linkage/homozygosity approach

Patient ID	Whole Exome Sequencing (Platform/ Manufacturer)	Homozygosity mapping/ Linkage analysis	
		(Platform/ Manufacturer)	Mapping software
A-II:1	Agilent SureSelect Human v2, SOLiD 5500xl Life Technologies, Lifescape v2.2	-	-
B-II:1	Agilent SureSelect Human v2, SOLiD 5500xl Life Technologies, Lifescape v2.3	-	-
C-II:2	Agilent SureSelect Human v5, Illumina HiSeq 2500	Affymetrix Genome-Wide Human SNP Array 6.0	AgileMultideogram [18]
D-II:1 and II:2	Nextera Expanded Exome Enrichment Kit and Illumina HiSeq2500 (30x coverage)	-	-
E-II:3	Agilent SureSelect Human v2, SOLiD 5500xl Life Technologies, Bioscope v2.1	-	-
F-II:2	Agilent SureSelect Human V4, Illumina HiSEQ 2000	Affymetrix Genome-Wide Human SNP Array 250K	Affymetrix Chromosome Analysis Suite (ChAS) [19]
G-III:4	Agilent SureSelect Human v2, SOLiD 5500xl Life Technologies, Lifescape v2.1	Illumina 6K SNPs array	Homozygosity mapper [20]
H-II:1	Agilent SureSelect Human v2, SOLiD 5500xl Life Technologies, Lifescape v2.6	-	-
I-II:1	Agilent SureSelect Human v4, SOLiD 5500xl Life Technologies	Affymetrix Genome-Wide Human SNP Array 6.0	Affymetrix Chromosome Analysis Suite (ChAS) [19]
J-III-3	Agilent SureSelect Human V4, Illumina HiSEQ 2000	Affymetrix Genome-Wide Human SNP Array 250K	Affymetrix Chromosome Analysis Suite (ChAS) [19]
K-II:1	Agilent SureSelect Human v2, SOLiD 5500xl Life Technologies, Lifescape v2.4	-	-

Supplemental Figure S1: Amino acid conservation for the missense variants identified in FDFT1 in Human and Baker's yeast.

