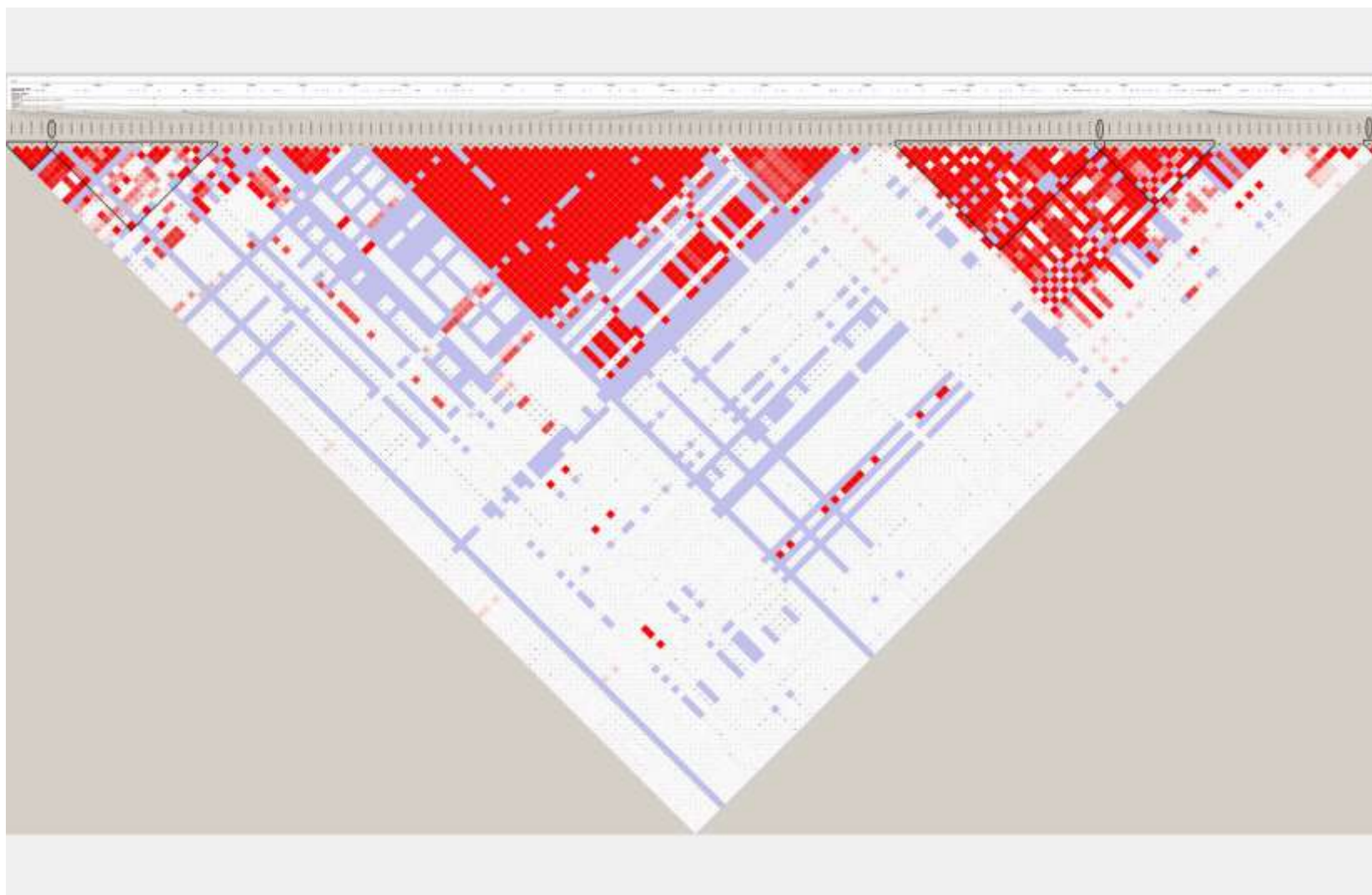


# **SUPPLEMENTAL MATERIAL**

**Figure S1. LD structure and haplotype block in *FGF12B* in Chinese Han population.** Linkage disequilibrium ( $D'$ ) for SNPs spanning a 266.3kbps genomic region in *FGF12B* on chromosome 3, are generated by Haploview 4.0.



**Figure S2. A schematic giving the standard, stylized intron/exon gene map of FGF12. Three tag SNPs and area covered by the disequilibrium blocks they tag, the three new rare mutations, and the three common SNPs identified in patients by the re-sequencing were indicated on the schematic.**

**a**



**b**



**c**

718 tccccgcgac tgcctt

Model ID	Model name	Score	Relative score	Start	End	Strand	predicted site sequence
MA0956.1	MZF1	6.510	0.573751908405984	1	6	-1	cggggA
MA0632.1	TcfE	6.021	0.575448175861752	1	10	-1	gtcgcgggA
MA0632.1	TcfE	6.021	0.675448175861752	1	10	1	Tccccgcgac
MA0831.1	TFE3	4.623	0.815167614162939	1	18	1	Tccccgcgac
MA0007.1	Pax2	6.425	0.89629859110259	4	11	-1	agtcgag
MA0498.2	MEIS1	0.901	0.800468963468584	6	12	1	gtactg

**d**

718 tccccacgac tgcctt

Model ID	Model name	Score	Relative score	Start	End	Strand	predicted site sequence
MA0956.1	MZF1	9.095	0.995687318791103	1	6	-1	tggggA
MA0130.1	ZNF354C	6.723	0.904024650450988	2	7	1	cggcAc
MA0259.1	ARNT-HPIA	4.644	0.98447244204401	4	11	-1	agtcgag
MA0007.1	Pax2	5.855	0.872915401603088	4	11	-1	agtcgag
MA0009.1	MFG-TFEZL1	4.209	0.805692934199387	6	10	1	cacgac
MA0498.2	MEIS1	2.103	0.927618881003162	6	12	1	agactg

**Figure S3. Transcription Factor MZF1 and ZNF354C do not regulate the expression of the FGF12 at the mutant site c.G723A.** **a**, Effect of MZF1 on the pGL3-Basic-FGF12B-5'UTR-Mut luciferase reporters compared with pGL3-Basic-FGF12B-5'UTR-Wt transfected into HeLa cells. **b**, Effects of ZNF354c on the pGL3-Basic-FGF12B-5'UTR-Mut luciferase reporters compared with pGL3-Basic-FGF12B-5'UTR-Wt. Luciferase activities were calculated as the ratio of firefly/renilla activities and normalized to the negative control (empty-vectors) group. Results were obtained from three independent experiments. Data are shown as means $\pm$ SD.

