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TABLE E1. Clinical information for individuals with reported disease causing *NOTCH1* mutations

Study	Mutation	ID	Gender	Age	Valve morphology	Cusp fusion	Aortic stenosis	Aortic insufficiency	Calc	AscAA, surgery (age in y), other CV abnormalities
Garg et al.	R1108X	II-1	F		TAV	N/A	+	+	+	Dilated AscAo, AVR (65)
	"*	II-2	F		BAV	Unk	+	?	+	AVR (76)
	"	III-3	F		Dysmorphic TAV	N/A	?	?	+	
	"*	III-5	M		BAV	Unk	Severe	?	+	Dilated AscAo, AVR (26)
	"	III-6	M		BAV	Unk	?	Severe	+	AVR (33)
	"	III-8	F		BAV	Unk	?	Mild	Unk	
	"	IV-2	M		TAV	N/A	Mild	?	+	
	"	IV-4	M		BAV	Unk	?	?	Unk	VSD, MS, parachute MV
	"	V-1	M		BAV	Unk	Mild	?	+	Dilated AscAo
	H1505del	II-1	F		BAV	Unk	Severe	?	+	AoR, AVR
	"	III-1	M		BAV	Unk	?	?	Unk	*MA, HLV, DORV*
	"	III-2	M		BAV	Unk	Severe	?	+	AVR
Mohamed et al.	T596M		M	49	BAV	R-L, R-non	Mild	Mild	+	Dilated AscAo [4.7 cm]
	P179H		M	55	BAV	R-L	Mild	Mild	+	Dilated AscAo [5.3 cm]
McKellar et al.	A1343V		F	41	BAV with Raphé	R-L	+	?	−	AVR, graft replacement of AscAo [at 4.3 cm]
	P1390T		M	55	BAV	R-non	+	?	+	AVR, reduction aortoplasty [at 4.5 cm]
Current study	T1090S	TII:4	M	56	TAV	N/A	−	−	+	Dilated AscAo [4.53 cm], abnormal diastolic function

ID, Identification; *Calc*, calcification; *AscAA*, ascending aortic aneurysm; *CV*, cardiovascular; *F*, female; *TAV*, tricuspid aortic valve; *N/A*, not applicable; *AscAo,* ascending aorta; *AVR*, aortic valve repair; *BAV*, bicuspid aortic valve; *Unk,* unknown; *M*, male; *VSD*, ventral septal defect; *MS*, mitral stenosis; *MV*, mitral valve; *AoR,* aortic root replacement; *MA*, mitral atresia; *HLV*; hypoplastic left ventricle; *DORV*, double outlet right ventricle; *R-L*, right-left commissural fusion; *R-non*, right-non commissural fusion. *Carriers assumed to have *NOTCH1* mutations but this was not confirmed by sequencing.

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000 Genotype–phenotype correlation in patients with bicuspid aortic valve and aneurysm

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Upon sequencing individuals from 13 families with ascending aortic aneurysm and bicuspid aortic valve, we conclude that *NOTCH1* mutations are rare and specifically relate to bicuspid aortic valve with prominent valve calcification and/or dysfunction. This apparent phenotype–genotype correlation may facilitate patient counseling and management.