Supplementary Material: Table S2

Table S2: Number and distribution of pathogenic variants (reported in literature, reported in Ensemble and gnomAD databases and total) per exon or intron of each MADD-related gene

Gene	Status	Number of pathogenic variants																							
		Exon													Intron										
		1	**2**	**3**	**4**	**5**	**6**	**7**	**8**	**9**	**10**	**11**	**12**	**13**	**1**	**2**	**3**	**4**	**5**	**6**	**7**	**8**	**9**	**10**	**11**	**12**	**13**
ETFA	**Literature**	2	3	2	3	4	**6**	3	0	5	3	3	0		0	0	0	0	0	**1**	**1**	0	0	0	0	0	
	Databases	0	1	1	0	1	0	0	1	**4**	2	1	0		0	0	0	0	0	0	0	0	0	0	0	0	
	Total	2	4	3	3	5	6	3	1	**9**	5	4	0		0	0	0	0	0	**1**	**1**	0	0	0	0	0	
ETFB	**Literature**	2	**5**	1	2	4	1								0	1	0	1	**2**	0							
	Databases	3	1	3	1	**5**	1								0	0	0	0	0	0							
	Total	5	6	4	3	**9**	2								0	1	0	1	**2**	0							
ETFDH	**Literature***	3	20	**33**	10	14	7	16	16	21	22	29	22	27	2	2	**4**	2	3	2	3	0	1	2	2	2	0
	Databases	0	0	4	1	4	0	3	8	5	2	6	**10**	5	0	0	0	0	0	0	0	0	0	0	0	0	0
	Total	3	20	**37**	11	18	7	19	24	26	24	35	32	32	2	2	**4**	2	3	2	3	0	1	2	2	2	0
FLAD1	**Literature**	1	**9**	0	0	1	1	0							0	0	0	0	**1**	0	0						
	Databases	3	**6**	5	0	5	3	1							0	0	0	0	0	0	0						
	Total	4	**15**	5	0	6	4	1							0	0	0	0	**1**	0	0						
SLC25A32	**Literature***	0	0	0	**2**	0	0	0							0	0	0	0	0	0	0						
	Databases	5	3	2	5	1	**9**	3							0	0	0	0	0	0	0						
	Total	5	3	2	7	1	**9**	3							0	0	0	0	0	0	0						
SLC52A1	**Literature***	0	0	0	0	0									0	0	0	**1**	0								
	Databases	0	0	0	0	0									0	0	0	0	0								
	Total	0	0	0	0	0									0	0	0	**1**	0								
SLC52A2	**Literature**	1	4	**19**	4	0									0	0	0	0	0								
	Databases	2	2	**5**	2	0									0	0	0	0	0								
	Total	3	6	**24**	6	0									0	0	0	0	0								
SLC52A3	**Literature***	0	**21**	10	7	12									0	0	0	**1**	0								
	Databases	0	4	7	1	**12**									0	0	0	0	0								
	Total	0	**25**	17	8	24									0	0	0	**1**	0								

* Variants that are located in the 5‘UTR and 3’UTR or not restricted to a single exon/intron have been omitted from Table S2. This incudes c.-75A>G, c.1989A>C, exon 1-5del, exon 1-7del and exon 7-8del in *ETFDH*, c.-264_31delins(14) in *SLC25A32*, the 1.9kb deletion spanning exons 2 and 3 of *SLC52A1* and c.1555C>T in *SLC52A3;* Bold values signify the exon and intron containing the largest number of variants per gene.