Additional file 1. ACDMPV patients enrolled in transcriptomic studies.

Patient / Sample	Lab Number	Group (Molecular defect at chr16q24.1)	Coordinates (hg38)	Parental origin of the chromosome 16, carrying pathogenic variant	Reference
C1	C1	Controls	N/A	N/A	N/A
C3	C3		N/A	N/A	N/A
C4	C4		N/A	N/A	N/A
1.1	60.4	1 (Enhancer deletion)	chr16:83,639,777/871-86,264,678/772	Maternal	Szafranski et al. 2013
1.2	28.7		~chr16:86,106,893-86,251,893	Maternal	Stankiewicz et al. 2009
1.3	155.3		chr16:84,457,588/632-86,238,392/436	Maternal	Szafranski et al. 2018
1.4	179.3		chr16:83,637,918/969-86,262,821/872	Paternal	Szafranski et al. 2018
2.1	90.3	2 (Enhancer + *FOXF1* + *FENDRR* deletion)	chr16:81,061,049-86,603,147	Maternal	Sen et al. 2013
2.2	118.3		chr16:85,993,409/427-87,165,121/139	Maternal	Prothro et al. 2016
2.3	170.3		chr16:85,030,576-86,687,561	Maternal	Szafranski et al. 2018
3.1	115.3	3 (*FOXF1* + *FENDRR* deletion)	chr16:86,471,844-86,541,855	Maternal	Szafranski et al. 2016
3.2	176.3		~chr16:86,508,525-86,515,660	Maternal	This work
4.1	73.3	4 (*FOXF1* mutation)	NM_001451.3:c.302C>T p.Ser101Leu	-	Sen et al. 2013
4.2	123.3		NM_001451.3:c.849_850del p.Ile285Glnfs*9	-	Szafranski et al. 2016
5.1	20.8	5 (Etiology unknown)	N/A	N/A	This work

N/A, not applicable; -, data not available.

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