

Supplementary Table S1: Publications reporting of CMMR-D patients

Authors, year	PMID no.	no. families	no. patients	patients also published by	PMID #
Ricciardone et al., 1999	9927033	1	3	Alotaibi et al., 2008	17889038
Wang et al., 1999	9927034	1	2		
Miyaki et al., 1997	9419979	1	1		
Hamilton et al., 1995,	7661930	1	2	Taylor et al., 1999, De Vos et al., 2004	10223463 15077197
De Rosa et al., 2000	10763829	1	2		
Trimbath et al., 2001	14574005	1	3	Shalon et al., 1997	9252914
Vilkki et al., 2001	11389087	1	1		
Whiteside et al., 2002	11809679	1	1		
Bougeard et al., 2003	12549480	1	2		
Raevaara et al., 2004	15139004	1	1		
Gallinger et al., 2004	14762794	1	3	Durno et al., 2005 Durno et al., 2011 Holter et al., 2011	15845562 22180144 21182953
Rey et al., 2004	15571801	1	1		
De Vos et al., 2004	15077197	1	4	Durno et al., 2005,	15845562
Menko et al., 2004	15340263	1	1		
Hegde et al., 2005	16000562	1	2		
Ostergaard et al., 2005	16283678	1	2		
Agostini et al., 2005	16144131	1	1		
Müller et al., 2006	16372347	1	2		
Marcos et al., 2006	16769400	1	1		
			10		
De Vos et al., 2006	16507833	5	(1+2+3+3+1)		
Plaschke et al., 2006	16418736	2	2 (1+1)		
Scott et al., 2007b	17259933	1	1		
Will et al., 2007	17258725	1	1		
Auclair et al., 2007	17557300	2	4(2+2)		
Poley et al., 2007	17440981	1	1	Wagner et al., 2003, Fam Cancer 2(Suppl 1):57	
Scott et al., 2007a	17601929	1	3		
Toledano et al., 2008	19101824	1	2		
Kratz et al., 2008	18007577	1	2		
Etzler et al., 2008	18030674	2	3 (2+1)		
Rahner et al., 2008	18409202	1	1		
Krüger et al., 2008	17851451	2	6 (4+2)	Gottschling et al., 2008	17851451
Gururangan et al., 2008	17993636	1	2		
Tan et al., 2008	18376293	1	2		
Jackson et al., 2008	18273873	1	1	Holter et al., 2011	21182953

Senter et al., 2008	18602922	5	5 (1+1+1+1+1)	Roy et al., 2009	19283792
Sjursen et al., 2008	19039682	1	2		
Kets et al., 2009	18781192	1	2		
Kratz et al. 2009	19293170	1	1		
Giunti et al., 2009	19156169	2	5 (2+3)		
Peters et al. 2009	19194194	1	1		
Ripperger et al., 2010	20015892	1	1		
Vaughn et al. 2010	20205264	5	5 (1+1+1+1+1)		
Jaspersen et al. 2010	21039432	1	2		
Holter et al., 2011	21182953	1	1		
Ilencikova et al., 2011	21674763	1	3		
Vaughn et al. 2011	21618646	1	1		
Plon et al., 2011	21356188	1	2		
Herkert et al., 2011	21376568	4	5 (1+2+1+1)		
Johannesma et al., 2011	21261604	1	2		
Leenen et al. 2011	21204794	1	2		
Yeung et al. 2012	22848017	1	1		
Baas et al. 2012	22692065	3	3 (1+1+1)		
Vasovcak et al. 2012	22608206	1	1		
Furlanetti et al. 2012*	22837778	1	2		
Chmara et al. 2013	23629955	2	3 (1+2)		
Lindsay et al. 2013	23582141	1	1		
total		79	127		
Unpublished patients data collected by Austrian, French, Israeli and Spanish consortium members **					
		12	19		
total (published and unpublished cases)		91	146		

* diagnosis in this patient was confirmed by identification of a homozygous MSH6 germ line mutation (Wimmer & Furlanetti unpublished results)

Supplementary Table S2: Malignancies in 146 CMMR-D patients separated by genotype

Malignancies	Number of tumors	Median age at diagnosis in years	Age range at diagnosis in years	Number of tumors	Median age at diagnosis in years	Age range at diagnosis in years	Number of tumors	Median age at diagnosis in years	Age range at diagnosis in years	Number of tumors	Median age at diagnosis in years	Age range at diagnosis in years
	total			MLH1/MSH2			MSH6			PMS2		
Haematological malignancies												
non-Hodgkin's lymphoma	31	5	0.4 - 17	10	2.5	0.4 - 10	7	6	3 - 11	14	9	3 - 17
lymphoid leukemia	9	6	2 - 21	1	2		1	6		7	6	2 - 21
acute myeloid leukemia	5	9.5	6 - 17	2	2	2 & n.k.	2	9.5	9 & 10	1	17	
malignant lymphoma	1	15		0			0			1	15	
atypical chronic myeloid leukemia	1	1		1	1		0			0		
acute leukemia	1	2		1	2		0			0		
total	48	6	0.4 - 21	15	2,5	0.4 - 10	10	7	3 - 11	23	9	2 - 21
Malignant brain and central nervous system tumour												
high grade gliomas	58	9.5	2 - 40	10	8	3 - 14	14	9	2 - 16	34	10	2 - 40
sPNET	8	8	4 - 17	0			0			8	8	4 - 17
medulloblastoma	7	7	4 - 12	2	7	7 & n.k.	3	6	4 - 7	2	9.5	7 & 12
infiltrating cerebral angiosarcoma	1	2		0			0			1	2	
cerebral anaplastic glioma	1	9		0			0			1	9	
papillary ganglioneural tumor	1	11		0			0			1	11	
not specified brain tumor	5	8	4 - 24	0			0			5	8	4 - 24
total	81	9	2 - 40	12	7	3 - 14	17	9	2 - 16	52	9	2 - 40
LS-associated carcinomas												
colon/rectum *	59	16	8 - 48	12	13	9 - 48	8	14.5	8 - 31	39	16	9 - 32
duodenum/jejunum/ileum	18	28	11 - 42	3	16	11 - 39	0			15	30	15 - 42
endometrium	6	28	23 - 44	1	44		1	24		4	27.5	23 - 35
bladder/ureter (papillary transitional cells)	2	20	19 & 21	0			0			2	20	19 & 21
ureter/renal pelvis (unspecified)	2	19	15 & 22	0			0			2	19	15 & 22
ovaries	1	17		0			0			1	17	
total	88	17	8 - 48	16	14.5	9 - 48	9	16	8 - 31	63	19	9 - 42
Others												
neuroblastoma	1	13		0			0			1	13	
nephroblastoma (Wilms tumour)	1	4		1	4		0			0		
ovarian neuroectodermal tumour	1	21		0			0			1	21	
infantile myofibromatosis	1	1		0			0			1	1	
rhabdomyosarcoma	1	4		0			0			1	4	
basal cell carcinoma	1	n.r.		0			0			1	n.r.	
muco-epidermoid ca. of parotis	1	11		0			0			1	11	
osteosarcoma	1	24		0			0			1	24	
total	8			1			8			7		

* multiple synchronous bowel carcinomas were counted as one malignancy; n.k. = not known