Supplementary Table-S10: Allelic imbalance events of OA susceptibility SNPs in articular cartilage of OA patients

Study	OA type	risk SNP	EA	EAF	Risk gene	SNPs_EA Risk - AI	LD with risk SNP R^2	Alt Allele AI	Meta Phi AI	Pval AI	Ref Allele AI	Direction effect OA risk allele
[1]	Hip & Knee	rs12901499	G	0.57	*SMAD3*	rs12901499_G - rs8031440_G	0.01					AI observed but not related to risk SNP. Results identical to Raine et al. [2]
						rs8031440_G - s3743342_C	1.00	T	0.57	0.170	C	
						rs8031440_G - rs3743343_T	0.11	C	0.58	0.010	T	
[3, 4]	Hip	rs143383 = rs143384	T	0.74	*GDF5*	rs143383_T - rs143384_A	0.82	G	0.50	0.950	A	No AI with risk allele. In contrast to paper Reynard et al. [5]
[6]	Hip	rs288326	T	0.08	*FRZB*	rs288326_T - rs288326_T	NA	T	0.50	0.220	C	No AI detected with risk allele
		rs7775	C	0.11		rs7775_C	NA	C	0.48	0.000	G	AI detected, FRZB lower is risk. Different to Snelling et al. [7]
[8]	Hip	rs225014	C	0.34	*DIO2*	rs225014_C	NA	C	0.50	0.970	T	No AI detected with risk allele. In contrast to paper Bos et al.[9]
[10]	Hip	rs2615977	C	0.22	*COL11A1*	rs2615977_C - rs1031820_A	0.16	A	0.48	0.004	G	Possible AI detected, lower COL11A1 with risk allele, see also Raine et al. [11]
[12]	LDG	rs1676486	T	0.3		rs1676486_T	NA	C	0.51	0.202	T	AI detected, lower COL11A1 with risk allele similar to results Mio et al.[12] & Raine et al.[11]
						rs1676486_T - rs2229783_T	0.32	G	0.53	0.000	A	
[13]	Hip	rs4836732	C	0.47	*ASTN2*	ND						
					PAPPA	ND						
[13]	Hip & Knee	rs11177	T	0.38	*GNL3*	rs11177_T - rs11177_T	NA	T	0.47	0.290	C	No AI different to Gee et al. [14]
					SPCS1	rs11177_T - rs6617_G	0.94	G	0.51	0.330	C	No AI different to Gee et al. [14]
					GLT8D1	rs11177_T - rs6976_T	1.00	T	0.50	0.990	C	No AI similar to Gee et al.[14]
[13]	Knee	rs12107036	G	0.52	*TP63*	rs12107036_G	ND					
[13]	Knee	rs10947262	C	0.58	*BTNL2*	rs10947262_C	ND					
[13]	Hip & Knee	rs2277831	A	0.743	*MICAL3*	rs2277831_A - rs7675_T	0.03	T	0.56	0.240	C	AI detected double heterozygotes with risk SNP. *MICAL3* low is risk based on eQTL. Similar to Ratnayake et al.[15]

Study	**OA type**	**risk SNP**	**EA**	**EAF**	**Risk gene**	**SNPs_EA Risk - AI**	**LD with risk SNP R^2**	**Alt Allele AI**	**Meta Phi AI^1**	***P* AI**	**Ref Allele AI**	**Direction effect OA risk allele**
[13]	Knee	rs11280	G	0.285	*C6ORF130* *OARD1*	rs11280_G - rs11280_G	NA	G	0.48	0.550	A	No AI
[13]	Hip	rs9350591	T	0.11	*SENP6*	rs9350591_T - rs17414687_A	0.07	A	0.50	0.970	G	Some AI not specifically among double heterozygotes
					COL12A1	rs9350591_T - rs970547_T	0.01	T	0.53	0.001	C	AI detected not related to risk allele
						rs9350591_T - rs240736	0.03	G	0.48	0.607	A	
					MYO6	rs9350591_T - rs699186_T	0.06	C	0.45	0.180	T	AI detected not related to risk allele
					TMEM30A	rs9350591_T - rs41269315_G	0.20	A	0.53	0.110	G	Suggestive evidence for AI, *TMEM30A* low is risk
					FILIP1	ND						
					COX7A2	ND						
[13]	Hip & Knee	rs8044769	C	0.5	*FTO*	rs8044769 _C - rs708277_A	0.01	A	0.57	0.060	G	AI particularly in double heterozygotes rs8044769 & rs708277. *FTO* high is risk (eQTL)
[16]	mJSW Hip	rs10492367	T	0.19	*KLHDC5* *HLKL42*	rs10492367_T - rs9029_G	0.01	G	0.53	0.460	C	No AI detected
					PTHLH	ND						
[16]	mJSW	rs10948172	G	0.29	*SUPT3H*	ND						
			T	0.67	*CDC5L*	rs10948172_G - rs11572055_G	0.02	G	0.45	0.418	A	Some AI detected (low metaPhi) not related to risk allele
						rs10948172_G - rs2273666_A	0.01	A	0.37	0.049	T	
		rs12206662	A	0.07	*RUNX2*	rs12206662_A - rs62400377_G	0.01	G	0.47	0.170	A	Some allelic imbalance measured (low metaPhi), not related to risk allele as checked by double heterozygotes
[16]	mJSW	rs10471753	C	0.61	*PIK3R1*	rs10471753_C - rs66666989_T	0.01					AI detected for rs66666989 in *PIK3R1* possibly related to risk allele. Majority of double heterozygous show AI
						rs66666989_T - rs9291928_C	0.71	T	0.56	0.001	C	
						rs10471753_C - rs3756668_A	0.02	A	0.53	0.321	G	
						rs10471753_C - rs1043526_G	0.16	G	0.36	0.000	A	
[16]	mJSW (hip)	rs2862851	T	0.49	*TGFA*	ND						Risk allele TGFA lower expression based on eQTL.

Study	OA type	risk SNP	EA	EAF	Risk gene	SNPs_EA Risk - AI	LD with risk SNP R^2	Alt Allele AI	Meta Phi AI	*P* AI	Ref Allele AI	Direction effect OA risk allele
[16, 17]	Hip mJSW	rs12982744	C	0.38	*DOT1L*	ND						
[10, 16]	Hip, mJSW	rs835487	G	0.34	*CHST11*	rs835487_G - rs7847_T	0.01	T	0.50	0.880	C	No AI detected also not among double heterozygotes (N=2)
		rs11842874	A	0.93	*MCF2L*	ND						
[16, 18]	Hip	rs788748	A	0.49	*IGFBP3*	rs788748_A - rs6670_A	0.01	A	0.62	0.000	T	AI particularly in double heterozygotes rs788748 & rs6670. Lower expression is risk based on eQTL
						rs6670_A - rs10241749_A	0.33					
						rs6670_A - rs1027438	0.38					
[19]	Knee & Hand	rs3815148	G	0.23	*BCAP29*	rs3815148_G - rs2807_A	0.08	A	0.56	0.008	G	AI detected for rs2807 not related to risk allele
						rs3815148_G - rs2808_G	0.11	G	0.53	0.547	A	
						rs3815148_G - rs68170813	0.49	C	0.54	0.523	T	
					GRP22	ND						
					COG5	ND						
					HBP1	rs3815148_G - rs7794598_T	ND	T	0.51	0.840	C	No AI detected. Different to Raine et al.[20]
					PRKAR2B	ND						
		rs4730250	G	0.17	*DUS4L*	ND						
[21, 22]	sCOMP, Hip	rs10038	A	0.33	COMP	rs10038_A - rs4808155_G	0.07	G	0.38	0.000	A	AI detected, not correlated with risk allele
						rs10038_A - rs12327738_A	0.97	A	0.50	0.695	G	No AI detected with risk allele
[22]	Hip	rs6094710	A	0.04	*NCOA3*	ND						
[22]	Hip	rs5009270	A	0.3	*IFRD1*	rs5009270_A - rs2074796_A	0.26	A	0.47	0.461	G	No AI detected with risk allele
						rs5009270_A - rs225396_G	0.28	G	0.52	0.690	T	
[22]	Hip	rs3757837	C	0.06	*CAMK2B*	ND						
[23]	Hand	rs3204689	C	0.45	*ALDH1A2*	rs3204689_C		C	0.42	0.000	G	AI detected similar to Stykarsdottir et al.[23]
[24]	Hand	rs4764133	T	0.39	*MGP*	rs4764133_T - rs1800801_T	0.99	T	0.41	0.000	C	AI detected identical to den Hollander et al.[24]

[1] Phi expresses allele counts of the alternative allele relative to total allele counts (alternative + reference). ND = not determined, NS = not significant, NA = Not applicable

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