UNIVERSITY OF LEEDS

This is a repository copy of Sex specific associations in genome wide association analysis of renal cell carcinoma.

White Rose Research Online URL for this paper: http://eprints.whiterose.ac.uk/149165/

Version: Accepted Version

Article:

Laskar, RS, Muller, DC, Li, P et al. (105 more authors) (2019) Sex specific associations in genome wide association analysis of renal cell carcinoma. European Journal of Human Genetics, 27. pp. 1589-1598. ISSN 1018-4813

https://doi.org/10.1038/s41431-019-0455-9

© 2019, The Author(s), under exclusive licence to European Society of Human Genetics. This is an author produced version of an article published in European Journal of Human Genetics. Uploaded in accordance with the publisher's self-archiving policy.

Reuse

Items deposited in White Rose Research Online are protected by copyright, with all rights reserved unless indicated otherwise. They may be downloaded and/or printed for private study, or other acts as permitted by national copyright laws. The publisher or other rights holders may allow further reproduction and re-use of the full text version. This is indicated by the licence information on the White Rose Research Online record for the item.

Takedown

If you consider content in White Rose Research Online to be in breach of UK law, please notify us by emailing eprints@whiterose.ac.uk including the URL of the record and the reason for the withdrawal request.



eprints@whiterose.ac.uk https://eprints.whiterose.ac.uk/

	HGVS Nomenclature (GRCh37)	Nearest gene	Reference Allele##	Alternate Allele	Effect Allele [#]	Discovery				Replication				Combined					
SNP						Female (N=5,087)		Male		Female		Male		Female			Male		
								(N=8,143)		(N=2,974)		(N=8,113)		(N=8,061)			(16,256)		
						OR[95%CI]	Р	OR[95%CI]	Р		%CI] P	OR[95%CI] P	P	OR [95%CI]	Meta	Het	OR	Meta	Het
						01(00/001)	•						, 		P-value	Pvalue	[95%CI]	P-value	Pvalue
rs4903064	NC_000014.8:g.73279420T>C	DPF3**	Т	С	С	1.47	9.0x10 ⁻¹⁴	1.09 [1.01-1.19] 0.0	0.02	1.24	0.003	1.09 0.10	1.38	1 5×10-14	0.10	1.09	0.005 0	0.54	
						[1.33-1.62]			0.02	[1.07-1.42]		[0.98-1.21]	0.10	[1.27-1.50]	1.5X10	0.10	[1.03-1.16]	0.005 (0.54
rs2121266	NC_000002.11:g.46535924C>A	EPAS1**	С	А	С	1.06 [0.97-1.15]	0.20	1.2 [1.12-1.28]	2.5x10 ⁻⁷	1.14	0.03	1.15	0.000	1.09	0.02	0.001	1.18	1.8x10 ⁻⁹ 0	
										[1.00-1.30]		[1.05-1.26]	0.002	[1.01-1.17]			[1.12-1.25]		0.62
rs147304092	NC_000007.13:g.33442937A>G	BBS9*	A	G	G	0.62	0.0.406	⁶ 1.05 0.49	0.40	0.97		1.20		0.72	1.0x10 ⁻⁵ 0.03		1.11	0.08	
						[0.52-0.76]	2.0x10 ⁻ ⁰		0.49	[0.76-1.24] 0.82	0.82	[1.00-1.43] 0.05	0.05	[0.63-0.84]		0.03	[0.99-1.24]		0.54
rs13027293	NC_000002.11:g.120001118G>T	STEAP3*	G	Т	Т	1.23	8.0x10 ⁻⁶	0.96	96 -1.03] 0.28	0.87	0.87 0.04	1.00		1.10	0.01		0.97	0.37	0.52
								[0.89-1.03]		[0.77-1.00]		[0.91-1.10]	0.95	[1.02-1.19]		1.6x10⁻⁴	[0.92-1.03]		
rs6554676	NC_000005.9:g.1233350A>T	SLC6A18*	А	Т	А	0.62		0.81		0.80	+	0.81		0.68	1		0.81	i – – – – –	 I
						[0.51-0.77]	6.0x10 ⁻⁶	[0.69-0.95] 0.0	0.008	0.11	[0 67-0 99]	0.04	[0 58-0 81]	6.1x10 ⁻⁶	0.07	[0 72-92]	0.001	0.67	
rs10484683	NC_000006.11:g.148152077A>G	SAMD5**	A	G	G	0.96		0.8	4.7x10 ⁻⁷	1 04	0.63	0.07 0.00		0.00 0.01	0.68		0.83	1.7x10 ⁻⁸ (
						[0.30	0.48	0.0 [0 74-0 88]		1.04		0.35	0.20	0.30		2.9x10 ⁻⁴			0.20
										[0.09-1.21]		0.02-1.04		[0.90-1.07]	┟────┦		0.76-0.69]		
rs78971134	NC_000012.11:g.107652531A>G	BTBD11**	A	G	G	0.91	0.23	0.69	9] 4.5x10 ⁻⁸	0.97	0.78	0.87	0.10	0.10 0.93 0.93 0.10 0.93	0.26	0.21	0.75	1.6x10 ⁻⁸ (0.15
						[0.78-1.06]		[0.61-0.79]		[0.78-1.20]		[0.74-1.03]		[0.82-1.06]			[0.68-0.83]		 I

Table 1. Significant associations in the replication series and/or final joint meta-analysis

** indicates association identified at <5% FDR and * indicates association identified at <30% FDR in the discovery series;

OR[95%CI]= Odds Ratio [95% confidence interval]; Meta P-Value= Meta-analysis P-value; Het P-value= heterogeneity P-value among studies , HGVS=Human Genome Variation Society ## Reference and alternate alleles as given by dbSNP, # Allele for which the effect size was calculated