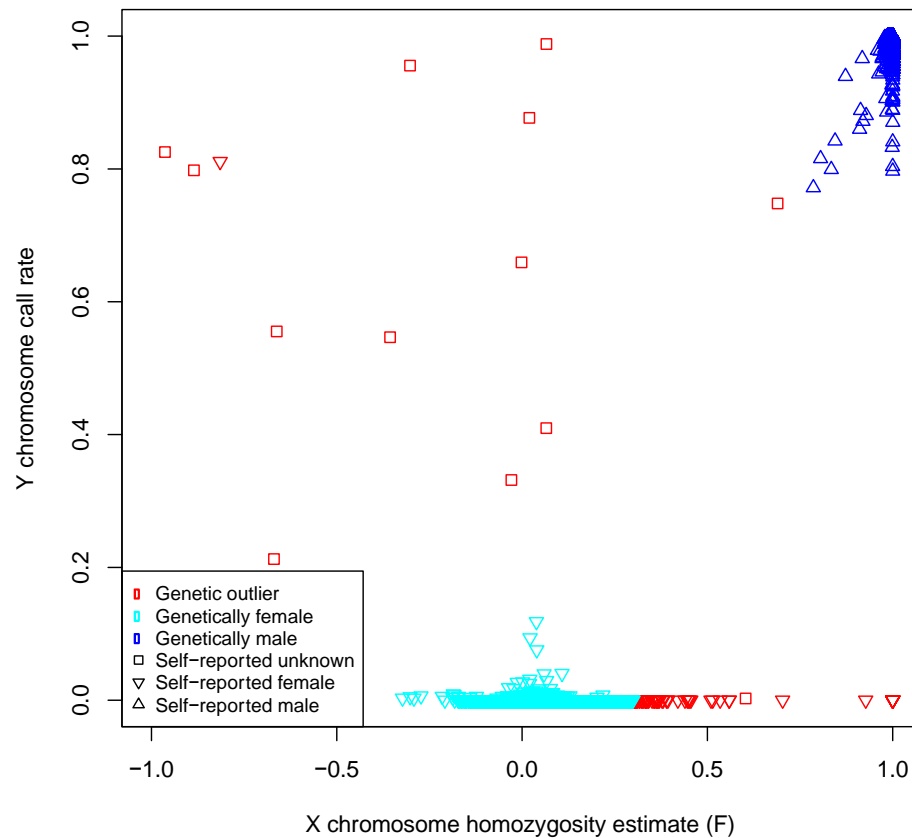


# Supplementary Notes and Figures

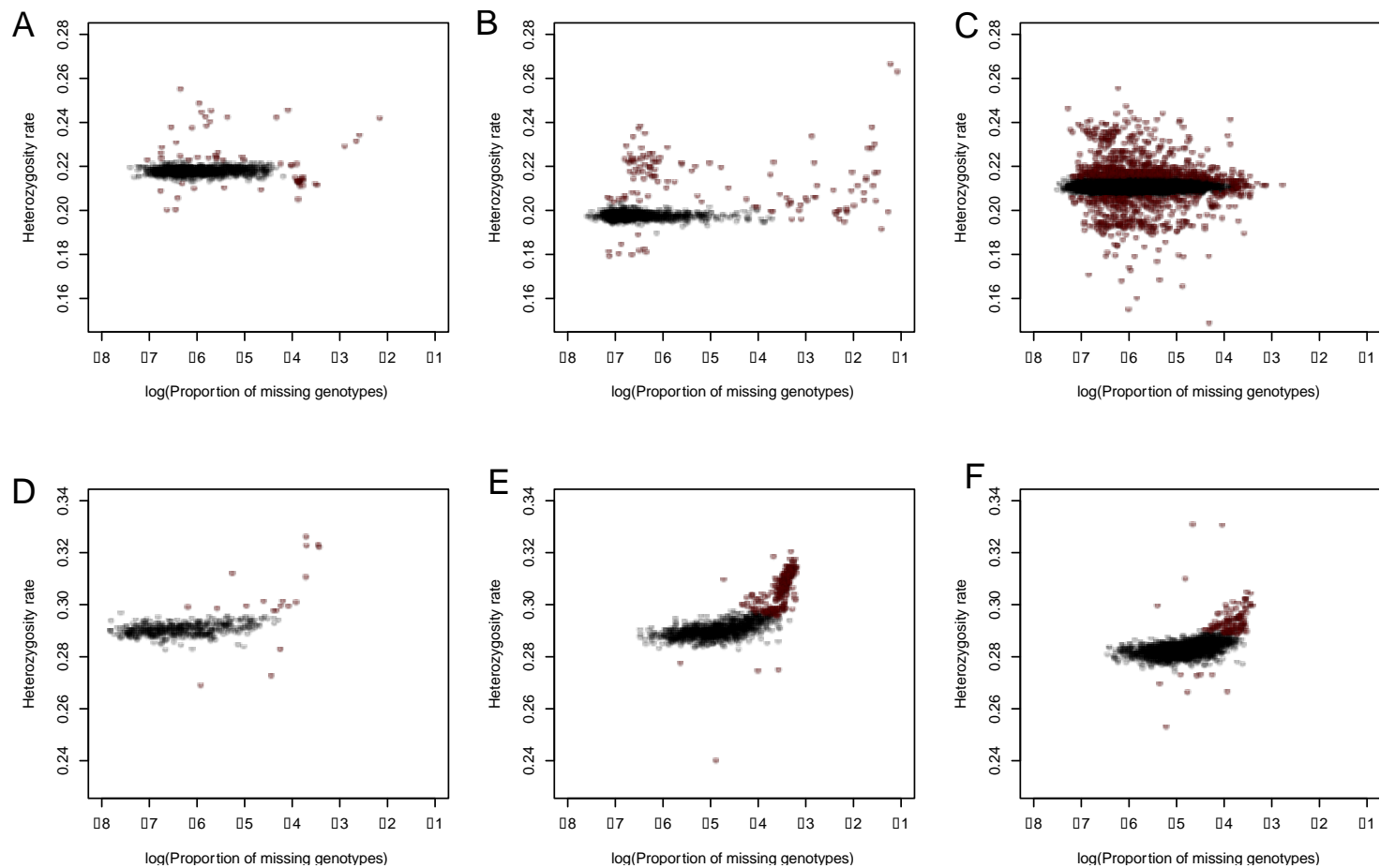
## The high comorbidity of inflammatory bowel disease in primary sclerosing cholangitis is only partly explained by shared genetic risk factors

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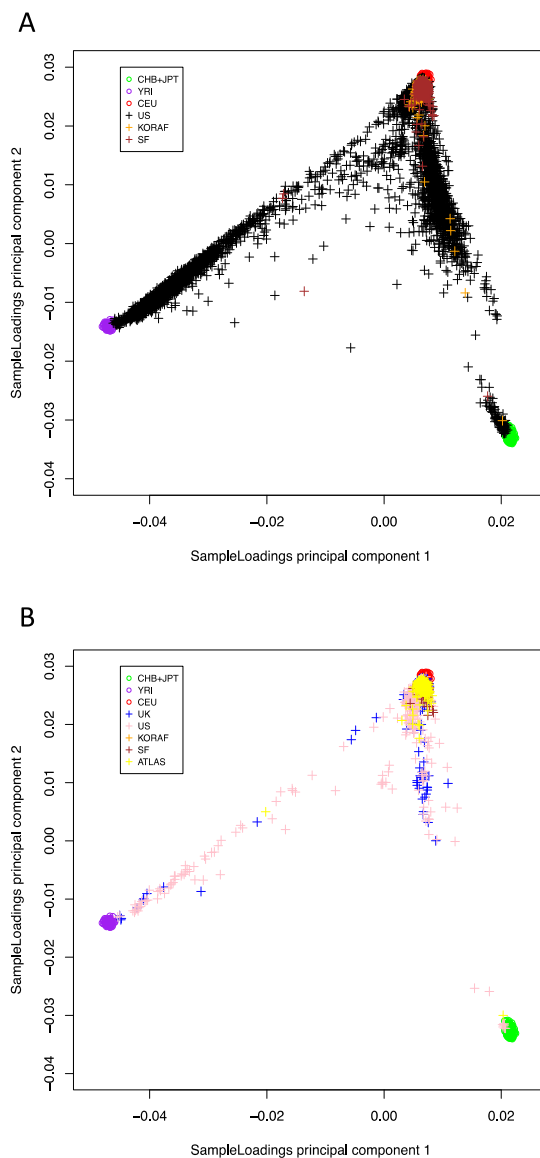
## Supplementary Figures



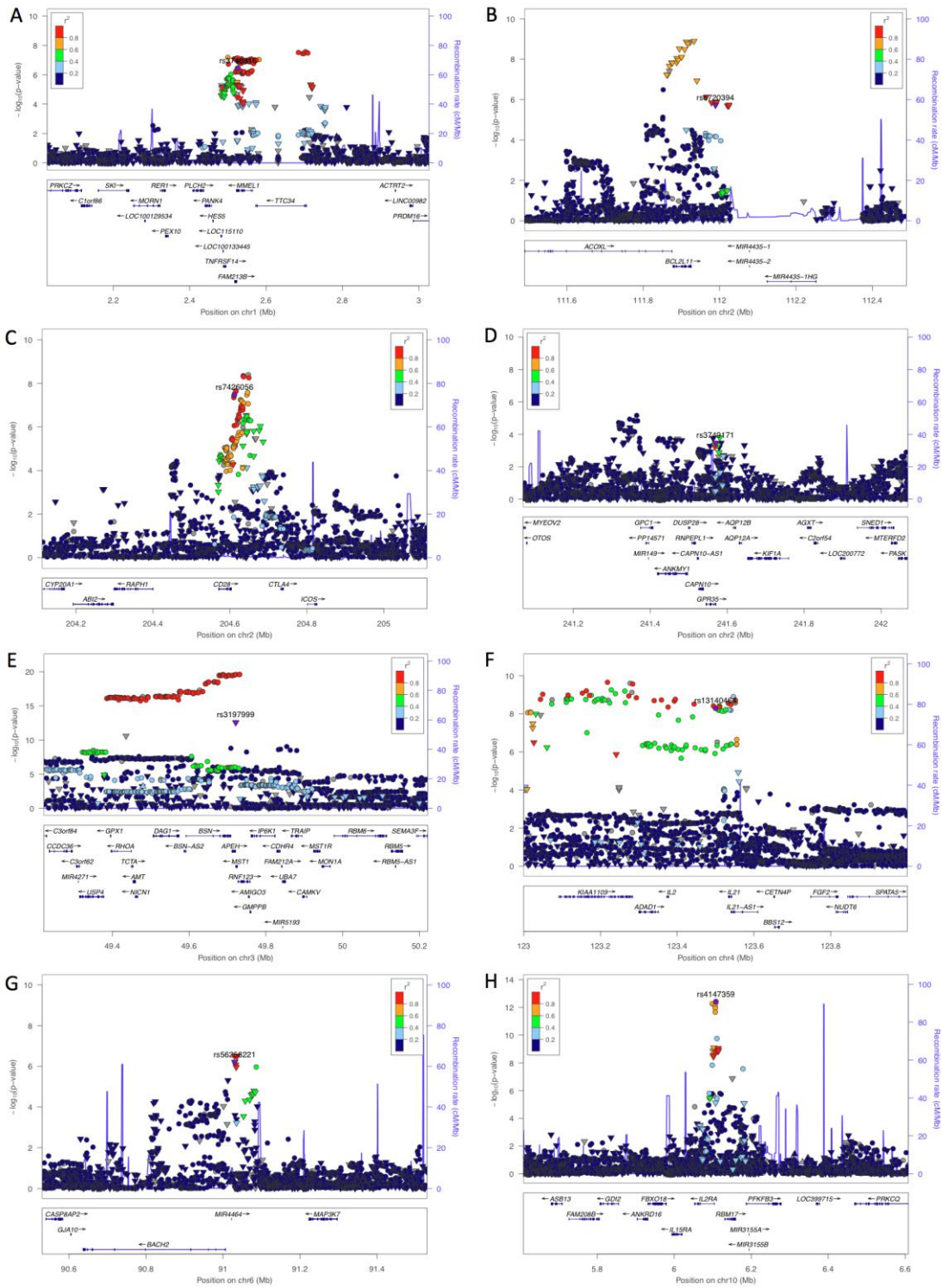
**Supplementary Figure 1. Ascertaining sex concordance and quality of genotyping from sex chromosome summary statistics.** Outlying individuals in this analysis are marked in red. Genetically defined sex was used in all downstream analyses.

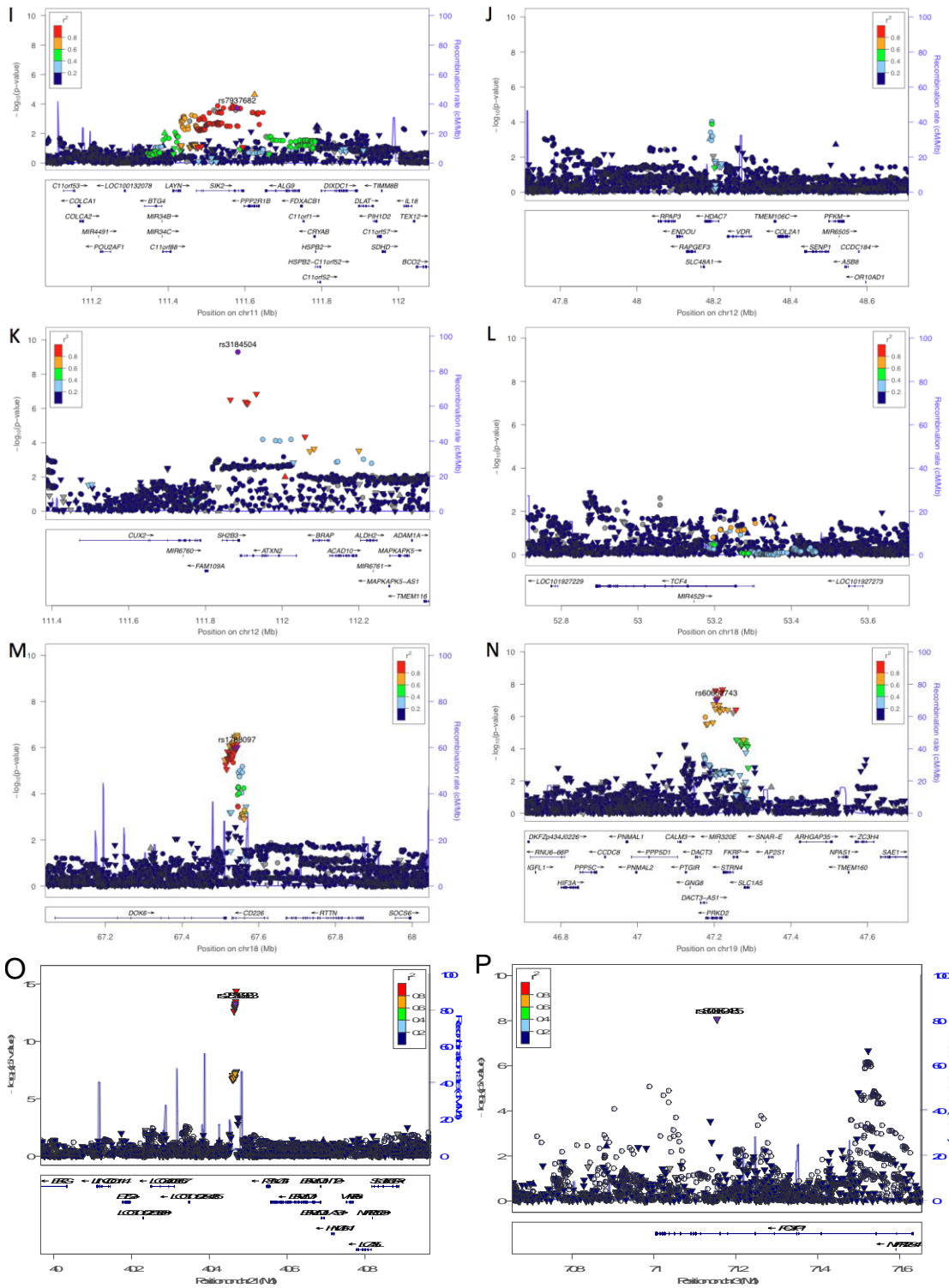


**Supplementary Figure 2. Outliers defined by heterozygosity rate and proportion of missing genotypes.** Outliers are coloured in red. A. All UK PSC cases, B. All US PSC cases, C. Non-African American samples in the HRS controls. D. All individuals genotyped in Berlin, E. All individuals genotyped in Munich, F. All individuals genotyped in San Francisco.

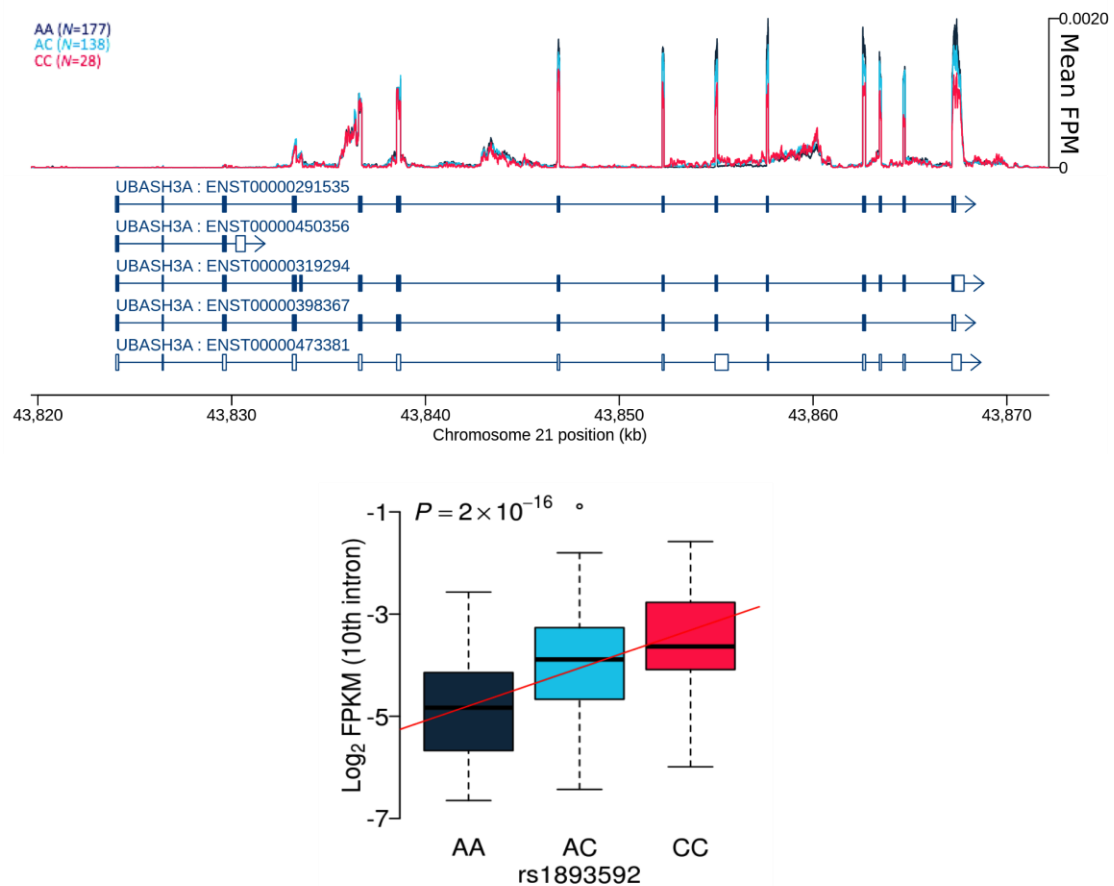


**Supplementary Figure 3. Individual ancestry inferred from PCA.** Red, purple and green circles are 1000 Genomes CEU (Utah residents with Northern and Western European ancestry), YRI (African), and CHB+JPT (Han Chinese and Japanese) A. All controls (crosses) overlaid onto the 1000 Genomes samples. B. All PSC cases (crosses) overlaid onto the 1000 Genomes samples. The number of non-European samples removed from this analysis is summarized in Supplementary Table 1.

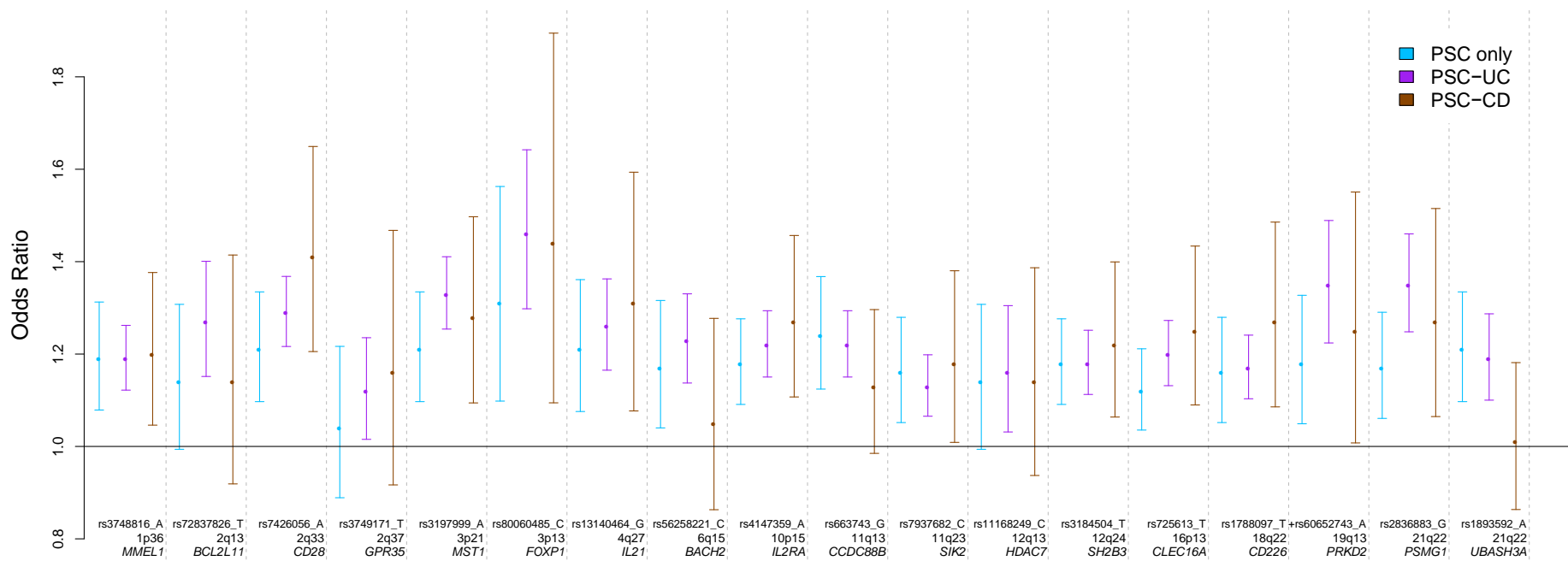




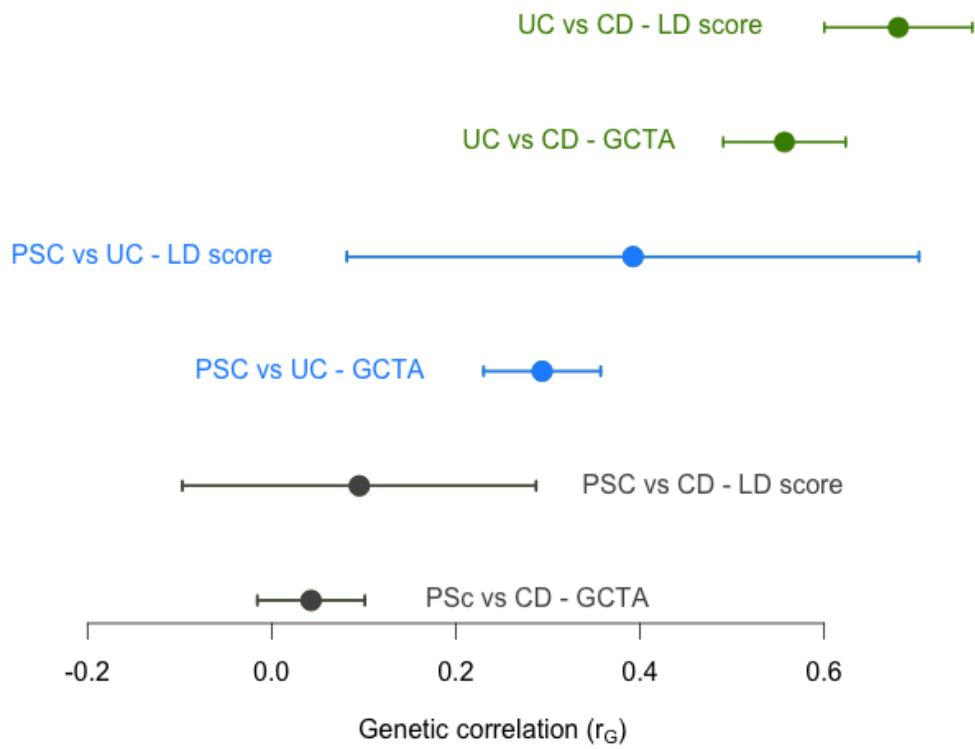




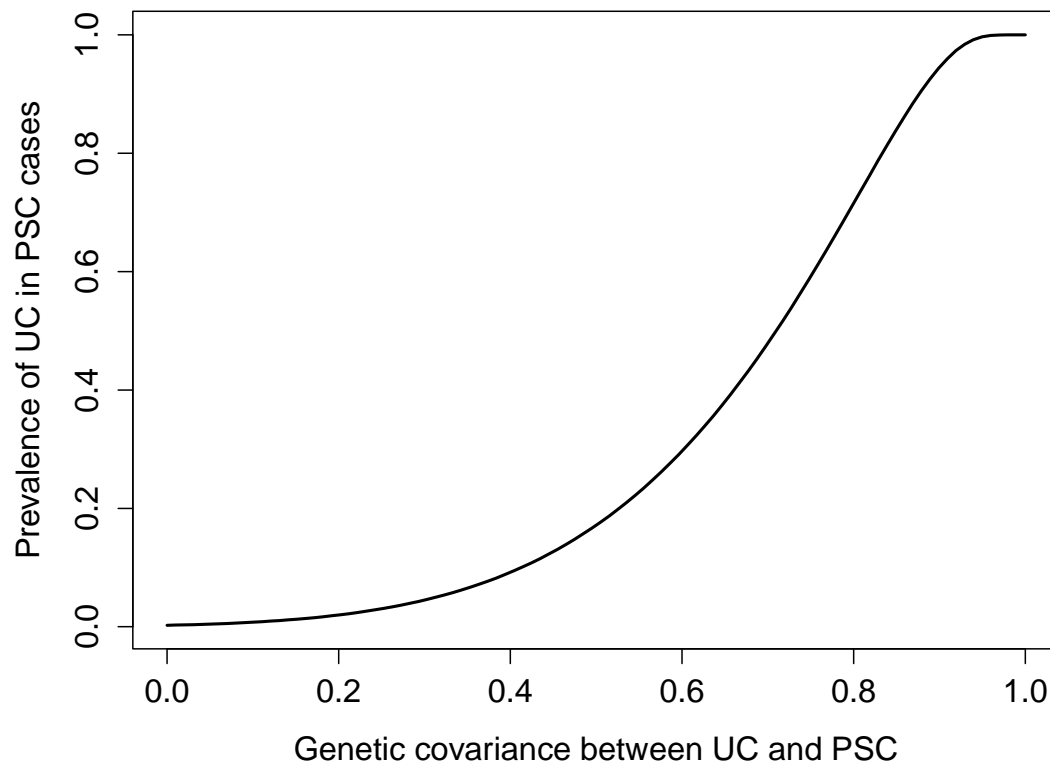
**Supplementary Figure 5. Effect of rs1893592 on the expression of intron 10 of *UBASH3A*.** The top panel shows the mean RNA-seq coverage around *UBASH3A* stratified by three different genotype groups at rs1893592. The bottom panel shows the boxplot of  $\log_2$  intron expression for the 10<sup>th</sup> intron against the three different genotype groups at rs1893592. FPM = (nucleotide count at the position)/(total nucleotide count for the sample)  $\times 10^6$ , FPKM = (fragment count + 1)/(cDNA length)/(total fragment count for the sample)  $\times 10^9$ . 343 European samples from the gEUVADIS project were used in this analysis



**Supplementary Figure 6. Odds-ratios of 18 PSC risk loci between different subsets of PSC patients.** Error bars represent 95% confidence intervals. No heterogeneity of odds could be observed between PSC subsets.



**Supplementary Figure 7. Consistency of co-heritability estimates between GCTA and LD score regression.** Point estimates of co-heritability and their 95% confidence intervals are plotted.



**Supplementary Figure 8. The effect of the genetic covariance of UC and PSC on the expected prevalence of UC in PSC cases.** For a given genetic covariance, the probability of having both disease is calculated under a bivariate liability model by numerically integrating the multivariate normal density assuming the prevalences of UC and PSC are 0.0025 and 0.0001, respectively.

## Supplementary Note

### Drug Identification

Protein-protein interactions (PPIs) for the twenty genes associated with PSC were identified using the DAVID, KEGG and BioCarta databases<sup>1-3</sup>. We then identified drugs in the drugbank database ([www.drugbank.ca](http://www.drugbank.ca)) that target these proteins and performed a literature search using PubMed ([www.pubmed.gov](http://www.pubmed.gov), last search March 9<sup>th</sup> 2015) and ClinicalTrials.gov ([www.clinicaltrials.gov](http://www.clinicaltrials.gov)) to assess which of these were the most promising for PSC. We highlight drugs that were 1) investigated for efficacy in PSC, 2) showed efficacy in other inflammatory disease or 3) deemed to have an interesting working mechanism. All drugs were selected based on evidence from phase I/II/III randomized clinical trials (RCTs) or published animal studies.

QC of the IBD and PSC GWAS cohorts for the genetic correlation analysis.

The genetic correlation analysis requires quality control of individual level genotype data to ensure that 1) the data are consistent across cohorts and diseases 2) there are no duplicate samples across phenotypes and 3) only the highest quality samples and genotypes are included in the analysis. Quality control of the PSC GWAS cohort is described in the Online Methods. Further QC to remove related individuals within the PSC GWAS cohort was conducted via PLINK<sup>4</sup> through pairwise identity by descent and the sample with lowest genotype call rate was removed for all pairs with identity by descent greater than 0.05. Quality control of the IBD GWAS data was conducted after merging all the 15 IBD cohorts and selecting autosomal SNPs that were well represented in all cohorts. Initially, SNPs out of Hardy-Weinberg equilibrium (HWE:  $P < 1 \times 10^{-6}$ ) in controls or with a call rate less than 90% were removed. Then an initial sample QC of removing individuals with call rate less than 95% was conducted. A set of 183,572 independent SNPs (pairwise  $r^2 < 0.1$ ) excluding a) the HLA region, b) SNPs with call rate  $< 98\%$ , c) significant deviation from HWE in controls ( $P < 1 \times 10^{-4}$ ), d) significant difference in call rate between cases and controls ( $P < 1 \times 10^{-6}$ ), and e)  $MAF < 10\%$  were used to estimate sample relatedness and ancestry. Pairwise identity by descent was estimated for all individuals that passed QC via PLINK, and the sample with the lowest genotype call rate was removed for all related/duplicated pairs

(IBD > 0.05). Sample ancestry was estimated via principal components analysis implemented in PLINK, whereby factor loadings from the 1000 Genomes were projected onto our cases and controls. Samples of non-European ancestry were inferred using Aberrant<sup>5</sup> and removed. The PSC and IBD post-QC genotypes were merged after removing triallelic SNPs or those with mismatching alleles. Sample QC on the combined data was conducted to identify duplicated and related samples. For this, 35,258 independent (pairwise  $r^2 < 0.1$ ) autosomal SNPs outside of the HLA were selected based on minor allele frequency (MAF > 10%) and genotype call rate (> 98%) in the combined dataset. Sample relatedness was measured by pairwise identity by descent and the sample with lowest genotype call rate was removed for all pairs with greater than 0.05 identity by descent. When duplicated samples were identified between PSC and IBD cohorts, the sample in the IBD cohort was removed to maximise power in the smaller PSC cohort. The number of samples removed by each QC step is summarized in Supplementary Table 13. 4,148,246 autosomal SNPs that were present throughout all the cohorts were used for this analysis.

#### Simulating heterogeneity of effect in cohorts of different sample size.

To further demonstrate that our analytical approach is robust to differences in sample size we simulated 1 million case and 1 million control genotypes at a SNP with an OR of 1.19 and a risk allele frequency (RAF) of 0.29. This represents the OR and RAF in UC at rs3197999 (*MST1*), a SNP that shows significant heterogeneity of odds versus PSC and significant evidence of causal variant colocalisation. Next, we randomly selected 4,796 case genotypes and 19,955 control genotypes from the respective pools of 1 million to represent the “PSC” cohort and 17,647 cases and 47,179 controls to represent the “UC” cohort. We repeated this selection process (with replacement) to create 1.2 million appropriately sized “PSC” and “UC” case-control cohorts under the null hypothesis of no heterogeneity of effect. We then used these simulated cohorts to ask the question “How many times under the null hypothesis of no significant heterogeneity of effect will sample size differences lead to significant evidence of heterogeneity?”. When the association P-value in the “PSC” cohort achieved genome-wide significance ( $P < 5 \times 10^{-8}$ ) we tested for association in the “UC” cohort and then tested for heterogeneity of odds. The P-value threshold for declaring significant evidence of heterogeneity was  $P < 2.78 \times 10^{-3}$ , to match that used in our study. An

empirical P-value was then derived that represents the proportion of simulations under the null that demonstrated significant evidence of heterogeneity of effect.

We repeated this simulation using the UC OR and RAF for each of the 4 SNPs that showed significant evidence of heterogeneity versus CD and UC and causal variant colocalisation. The results of this simulation study are shown in the table below.

rsID	Gene	UC OR	RAF	Empirical heterogeneity of odds P-value
rs3197999	<i>MST1</i>	1.19	0.29	$1.17 \times 10^{-4}$
rs13140464	<i>IL21</i>	1.12	0.84	$2.97 \times 10^{-4}$
rs1788097	<i>CD226</i>	1.07	0.5	$1.14 \times 10^{-4}$
rs3184504	<i>SH2B3</i>	1.05	0.49	$7.17 \times 10^{-5}$

Across the total 4.8 million simulations, only 68 ( $P=1.41 \times 10^{-5}$ ) resulted in a heterogeneity of odds P-value smaller than those we observed in our real data. This is highly significant for 4 statistical tests, and thus, these results clearly demonstrate that the sample size differences between the PSC and IBD cohorts cannot explain the significant degree of heterogeneity of effect we observe in our study.

1. Huang, D.W., Sherman, B.T. & Lempicki, R.A. Systematic and integrative analysis of large gene lists using DAVID bioinformatics resources. *Nature Protocols* 4, 44-57 (2009).
2. Huang, D.W., Sherman, B.T. & Lempicki, R.A. Bioinformatics enrichment tools: paths toward the comprehensive functional analysis of large gene lists. *Nucleic Acids Research* 37, 1-13 (2009).
3. Kanehisa, M. *et al.* Data, information, knowledge and principle: back to metabolism in KEGG. *Nucleic Acids Research* 42, D199-D205 (2014).
4. Purcell, S. *et al.* PLINK: a tool set for whole-genome association and population-based linkage analyses. *Am J Hum Genet* 81, 559-75 (2007).
5. Bellenguez, C. *et al.* A robust clustering algorithm for identifying problematic samples in genome-wide association studies. *Bioinformatics* 28, 134-5 (2012).

## List of Members in Contributing Consortia

### Members of the UK-PSC consortium

Abertawe Bro Morgannwg University NHS Trust (Dr. Chin Lye Ch'ng, Dr. Clement Lai, Dr. Tom Yapp), Aintree University Hospitals NHS Foundation Trust (Dr. Richard Sturgess), Airedale NHS Trust (Dr. Chris Healey), Barnsley Hospital NHS Foundation Trust (Dr. Kapil Kapur), Ashford and St Peter's Hospitals NHS Trust (Dr. John Thornton), Barking, Havering and Redbridge University Hospitals NHS Trust (Dr. Stephen Grainger, Dr. Purushothaman Premchand), Barnet and Chase Farm Hospitals NHS Trust (Dr. Stephen Mann, Dr. Kalpesh Besherdas), Barts and The London NHS Trust (Dr. Richard Marley, Prof Graham Foster), Basingstoke and North Hampshire NHS Foundation Trust (Dr. John Ramage), Bedford Hospital NHS Trust (Dr. Rory Harvey), Blackpool Teaching Hospitals NHS Foundation Trust (Dr Peter Isaacs), Bolton NHS Foundation Trust (Dr. George Lipscomb), Bradford Teaching Hospitals NHS Foundation Trust (Dr. Sulleman Moreea), Brighton and Sussex University Hospitals NHS Trust (Dr. Jeremy Tibble, Dr. Nick Parnell), Bromley Hospitals NHS Trust (Dr. Anthony Jenkins), Buckinghamshire Healthcare NHS Trust (Dr. Sue Cullen, Dr. David Gorard), Burton Hospitals NHS Foundation Trust (Dr. Altaf Palejwala), Cambridge University Hospitals NHS Foundation Trust (Dr. George Mells, Dr. Muhammad F Dawwas), Calderdale And Huddersfield NHS Foundation Trust (Dr. Susan Jones, Dr. Ashwin Verma), Cardiff and Vale NHS Trust (Dr. Richard Aspinall, Dr. Sunil Dolwani), Central Manchester University Hospitals NHS Foundation Trust (Dr. Martin Prince), Chelsea and Westminster Hospital NHS Foundation Trust (Dr. Karen Hawkins, Dr. Priyajit Prasad), Chesterfield Royal Hospitals NHS Foundation Trust (Dr. David Elphick), City Hospitals Sunderland NHS Foundation Trust (Dr. Harriet Mitchison), Colchester Hospital University NHS Foundation Trust (Dr. Ian Gooding), Countess of Chester Hospital NHS Foundation Trust (Dr. Mazn Karmo), County Durham and Darlington NHS Foundation Trust (Dr. Anjan Dhar, Dr. Stephen Mitchell, Dr. Sushma Saksena), Cwm Taf NHS Trust (Dr. Minesh Patel), Dartford and Gravesham NHS Trust (Dr. Roland Ede), Derby Hospitals NHS Foundation Trust (Dr. Andrew Austin), Doncaster and Bassetlaw Hospitals NHS Foundation Trust (Dr. Joanne Sayer), Dorset County Hospitals NHS Foundation Trust (Dr. Chris Hovell), East and North Hertfordshire NHS Trust (Dr. Martyn Carter, Dr. Peter McIntyre), East Cheshire NHS Trust (Dr. Konrad Koss), Medway NHS Foundation Trust (Dr. Gray Smith-Laing), Maidstone and Tunbridge Wells NHS Trust (Dr. George Bird), East Kent Hospitals University NHS

Foundation Trust (Dr. Kate Hill), East Sussex NHS Trust (Dr. David Neal), East Lancashire Hospitals NHS Trust (Dr. VishalKaushik), Epsom and St. Helier University Hospitals NHS Trust (Dr. Guan Lim), Frimley Park 57 NHS Foundation Trust (Dr. Aftab Ala), Gateshead Health NHS Foundation Trust (Dr. Athar Saeed), George Eliot Hospital NHS Trust (Dr. Sankara Raman), Gloucestershire Hospitals NHS Foundation Trust (Professor Jonathan Brown), Great Western Hospitals NHS Foundation Trust (Dr. Ajeya Shetty), Guy's and St Thomas' NHS Trust (Dr. Mark Wilkinson), Gwent Healthcare NHS Trust (Dr. Miles Allison, Dr. Marek Czajkowi, Dr. Peter Neville), Harrogate and District NHS Foundation Trust (Dr. Butt, Dr. Gareth Davies), Heart of England NHS Foundation Trust (Dr. Theodore Ngatchu), Hereford Hospitals NHS Trust (Dr. Rupert Ransford), Hinchingsbrooke Health Care NHS Trust (Dr. Richard Dickinson), Homerton University Hospital NHS Foundation Trust (Dr. Ray Shidrawi), Hull And East Yorkshire Hospitals NHS Trust (Dr. George Abouda), Hywel Dda Health Board NHS Trust (Dr. Faiz Ali, Dr. Mark Narain, Dr. Ian Rees, Dr. Imroz Salam), Imperial College Healthcare NHS Trust (Dr. Ashley Brown), Ipswich Hospital NHS Trust (Dr. Simon Williams), NHS Isle Of Wight (Dr. Christopher Sheen), James Paget University Hospitals NHS Foundation Trust (Dr. Matthew Williams), Kings College Hospital NHS Foundation Trust (Dr. Michael Heneghan), Kingston Hospital NHS Trust (Dr. Chris Rodrigues), Lancashire Teaching Hospitals NHS Foundation Trust (Dr. Phillip Shields), Leeds Teaching Hospitals NHS Trust (Dr. Mark Aldersley, Dr. Mervyn Davies, Dr. Charles Millson), Luton and Dunstable Hospital NHS Foundation Trust (Dr. Sambit Sen), Mid Cheshire Hospitals NHS Foundation Trust (Dr Kevin Yoong), Mid Essex Hospital Services NHS Trust (Dr. Cho Cho Khin), Mid Staffordshire NHS Foundation Trust (Dr. Pradip Singh), Mid Yorkshire Hospitals NHS Trust (Dr. Nurani Sivaramakrishnan), Milton Keynes Hospital NHS Foundation Trust (Dr. George MacFaul), Newham University Hospital NHS Trust (Dr. Matthew Guinane), NHS Ayrshire & Arran (Dr. Chris Gillen, Dr. James Rose, Dr. Amir Shah), NHS Borders (Dr. Chris Evans), NHS Dumfries & Galloway (Dr. Subrata Saha), NHS Fife (Dr. Sherzad Balta, Dr. John Wilson), NHS Forth Valley (Dr. Peter Bramley), NHS Grampian (Dr. Andrew Fraser), NHS Greater Glasgow and Clyde (Dr. Aidan Cahill, Dr. G. Curry, Dr. Shouren Datta, Dr. Ewan Forrest, Dr. Neil Jamieson, Dr. James McPeake, Professor Peter Mills, Dr. Judith Morris, Dr. Adrian Stanley), NHS Highland (Dr. Dara De Las Heras, Dr. Tim Shallcross), NHS Lanarkshire (Dr. Stuart Campbell, Dr. Richard Crofton, Dr. Andrzej Prach), NHS Lothian (Dr. Andrew Bathgate, Dr. Kevin Palmer), NHS Tayside (Dr. Alan Shepard, Dr. John Dillon), Norfolk and Norwich University Hospitals NHS Foundation Trust (Dr. Martin Phillips), North Bristol NHS Trust (Dr. Robert Przemioslo), North Cumbria University Hospitals NHS Trust (Dr. Babur Javaid, Dr. Chris McDonald), North Tees And Hartlepool NHS Foundation Trust (Dr. Jane Metcalf), North Wales NHS Trust (Dr. Paulose George, Dr. David Ramanaden), The North West London Hospitals NHS

Trust (Dr. Maxton Pitcher), North West Wales NHS Trust (Dr. Jaber Gasem, Dr. Richard Evans), Northampton General Hospital NHS Trust (Dr. Udi Shmueli), Northern Devon Healthcare NHS Trust (Dr. Andrew Davis), Northern Lincolnshire and Goole Hospitals NHS Foundation Trust (Dr. Prabhakar Mysore, Dr. Asifabbas Naqvi), Northumbria Healthcare NHS Trust (Dr. Mark Welfare), Nottingham University Hospitals NHS Trust (Dr. Steve Ryder), Pennine Acute Hospitals NHS Trust (Dr. Howard Klass, Dr. Jimmy Limdi, Dr. Bashir Rameh), Peterborough Stamford Hospitals NHS Foundation Trust (Dr. Mary Ninkovic), Plymouth Hospitals NHS Trust (Dr. Matthew Cramp), Poole Hospital NHS Foundation Trust (Dr. Nicholas Sharer), Portsmouth Hospitals NHS Trust (Dr. Patrick Goggin), Queen Mary's Sidcup NHS Trust (Dr. Howard Curtis), Royal Berkshire NHS Foundation Trust (Dr. Jonathan Booth), Royal Cornwall Hospitals NHS Trust (Dr. Hyder Hussaini), Royal Devon and Exeter NHS Foundation Trust (Dr. Reuben Ayres), Royal Free Hampstead NHS Trust (Dr. Douglas Thorburn), Royal Liverpool and Broadgreen University Hospitals NHS Trust (Dr. Martin Lombard), Royal Surrey County Hospital NHS Trust (Dr. Michelle Gallagher), Royal United Hospital Bath NHS Trust (Dr. Duncan Robertson), Salisbury NHS Foundation Trust (Dr. Sam Vyas), Sandwell and West Birmingham Hospitals NHS Trust (Dr. Saket Singhal), Scarborough And North East Yorkshire Health Care NHS Trust (Dr. Sathish Babu), Sheffield Teaching Hospitals NHS Foundation Trust (Dr. Dermot Gleeson), Sherwood Forest Hospitals NHS Foundation Trust (Dr. Sharat Misra), Shrewsbury and Telford Hospital NHS Trust (Dr. Jeff Butterworth), South Devon Healthcare NHS Trust (Dr. Keith George), South London Healthcare NHS Trust (Dr. Alastair McNair), South Tees Hospitals NHS Trust (Dr. Andrew Douglas), South Tyneside NHS Foundation Trust (Dr. Colin Rees), South Warwickshire General Hospitals NHS Trust (Dr. Jeremy Shearman), Southampton University Hospitals NHS Trust (Dr. Kate Nash, Dr. Mark Wright), Southend University Hospital NHS Foundation Trust (Dr. Gary Bray), Southport And Ormskirk Hospital NHS Trust (Dr. Graham Butcher), St. George's Healthcare NHS Trust (Dr. Daniel Forton), St. Helens and Knowsley Hospitals NHS Trust (Dr. John McLindon), Stockport NHS Foundation Trust (Dr. Debashis Das), Surrey and Sussex Healthcare NHS Trust (Dr. Gary Mackenzie, Dr. Azhar Ansari, Dr. Gregory Whatley), Taunton and Somerset NHS Foundation Trust (Dr. Stirling Pugh), Dudley Group of Hospitals NHS Trust (Dr. Neil Fisher), The Hillingdon Hospital NHS Trust (Dr. Deb Datta), The Lewisham Hospital NHS Trust (Dr. John Odonohue), The Newcastle upon Tyne Hospitals NHS Foundation Trust (Dr. Mark Hudson), The Princess Alexandra Hospital NHS Trust (Dr. Rosemary Phillips), The Queen Elizabeth Hospital King's Lynn NHS Trust (Dr. Andrew Douds), Rotherham NHS Foundation Trust (Dr. Barbara Hoeroldt), Royal Bournemouth and Christchurch Hospitals NHS Foundation Trust (Dr. Earl Williams), The Royal Wolverhampton Hospitals NHS Trust (Dr. Matthew Brookes), Trafford Healthcare NHS Trust (Dr. Chris Summerton), United Lincolnshire Hospitals NHS Trust (Dr.

Aravamuthan Sreedharan, Dr. Sanjiv Jain, Dr. Martin James), University College London Hospitals NHS Foundation Trust (Dr. Stephen Pereira), University Hospital Birmingham NHS Foundation Trust (Professor David Adams), University Hospital of North Staffordshire NHS Trust (Dr. Alison Brind), University Hospital of South Manchester NHS Foundation Trust (Dr. Gill Watt), University Hospitals Bristol NHS Foundation Trust (Dr. Fiona Gordon, Dr. Jim Portal), University Hospitals Coventry and Warwickshire NHS Trust (Dr. Esther Unitt), University Hospitals of Leicester NHS Trust (Dr. Allister Grant), Walsall Hospitals NHS Trust (Dr. Mark Cox), Warrington and Halton Hospitals NHS Foundation Trust (Dr. Subramaniam Ramakrishnan), West Hertfordshire Hospitals NHS Trust (Dr. Alistair King), West Suffolk Hospitals NHS Trust (Dr. Simon Whalley), Western Sussex Hospitals NHS Trust (Dr. Andy Li, Dr. Mohammed Rashid), Weston Area Health NHS Trust (Dr. Andrew Bell), Winchester and Eastleigh Healthcare NHS Trust (Dr. Harriet Gordon), Wirral University Teaching Hospital NHS Foundation Trust (Dr. Riyaz Faizallah), Worcestershire Acute Hospitals NHS Trust (Dr. Ishfaq Ahmad, Dr. Ian Gee), Wrightington, Wigan and Leigh NHS Trust (Dr. Gurvinder Banait), Yeovil District Hospital NHS Foundation Trust (Dr. Steve Gore, Dr. James Gotto), York Hospitals NHS Foundation Trust (Dr. Alastair Turnbull).

## Members of the International IBD Genetics Consortium

Clara Abraham<sup>1</sup>, Jean-Paul Achkar<sup>2,3</sup>, Tariq Ahmad<sup>4</sup>, Leila Amininejad<sup>5,6</sup>, Ashwin N. Ananthakrish<sup>7,8</sup>, Vibeke Andersen<sup>9,10</sup>, Carl A Anderson<sup>11</sup>, Jane M. Andrews<sup>12</sup>, Vito Annese<sup>13,14</sup>, Gilles Jobin<sup>15,16</sup>, Leonard Baidoo<sup>17</sup>, Robert N Baldassano<sup>18</sup>, Tobias Balschun<sup>19</sup>, Peter A. Bampton<sup>20</sup>, Murray Barclay<sup>21</sup>, Jeffrey C Barrett<sup>11</sup>, Theodore M Bayless<sup>22</sup>, Johannes Bethge<sup>23</sup>, Joshua C Bis<sup>24</sup>, Alain Bitton<sup>25</sup>, Gabrielle Boucher<sup>26</sup>, Stephan Brand<sup>27</sup>, Steven R. Brant<sup>28</sup>, Carsten Büning<sup>29</sup>, Angela Chew<sup>30,31</sup>, Judy H Cho<sup>32</sup>, Isabelle Cleynen<sup>11,33</sup>, Ariella Cohain<sup>34</sup>, Anthony Croft<sup>35</sup>, Mark J. Daly<sup>36,37</sup>, Mauro D'Amato<sup>38</sup>, Silvio Danese<sup>39</sup>, Dirk DeJong<sup>40</sup>, Martine DeVos<sup>41</sup>, Goda Denapiene<sup>42</sup>, Lee A Denson<sup>43</sup>, Kathy L Devaney<sup>7</sup>, Olivier Dewit<sup>44</sup>, Renata D'Inca<sup>45</sup>, Marla Dubinsky<sup>46</sup>, Richard H Duerr<sup>47,48</sup>, Cathryn Edwards<sup>49</sup>, Jonah Essers<sup>50,51</sup>, Lynnette R. Ferguson<sup>52</sup>, Eleonora A Festen<sup>53</sup>, Philip Fleshner<sup>54</sup>, , Denis Franchimont<sup>5,6</sup>, Andre Franke<sup>19</sup>, Karin Fransen<sup>55</sup>, Richard Geary<sup>21,56</sup>, Michel Georges<sup>57,58</sup>, Christian Gieger<sup>59</sup>, Jürgen Glas<sup>25</sup>, Philippe Goyette<sup>26</sup>, Todd Green<sup>50,37</sup>, Anne M Griffiths<sup>60</sup>, Stephen L Guthery<sup>61</sup>, Hakon Hakonarson<sup>18</sup>, Jonas Halfvarson<sup>62</sup>, Katherine Hanigan<sup>35</sup>, Talin Haritunians<sup>54</sup>, Ailsa Hart<sup>63</sup>, Chris Hawkey<sup>64</sup>, Grant W Montgomery<sup>65</sup>, Matija Hedl<sup>66</sup>, Paul Henderson<sup>67,68</sup>, Xinli Hu<sup>69</sup>, Hailiang Huang<sup>36,37</sup>, Ken Y. Hui<sup>32</sup>, Marcin Imielinski<sup>18</sup>, Andrew Ippoliti<sup>54</sup>, Laimas Jonaitis<sup>70</sup>, Luke Jostins<sup>71,72</sup>, Tom H. Karlsen<sup>73,74,75</sup>, Nicholas A. Kennedy<sup>76</sup>, Mohammed Azam Khan<sup>77,78</sup>, Gediminas Kiudelis<sup>70</sup>, Krupa Krishnaprasad<sup>35</sup>, Subra Kugathasan<sup>79</sup>, Limas Kupcinskas<sup>80</sup>, Anna Latiano<sup>13</sup>, Debby Laukens<sup>41</sup>, Ian C. Lawrance<sup>31</sup>, James C. Lee<sup>81</sup>, Charlie W. Lees<sup>76</sup>, Marcis Leja<sup>82</sup>, Johan Van Limbergen<sup>83</sup>, Paolo Lionetti<sup>84</sup>, Jimmy Z. Liu<sup>11</sup>, Edouard Louis<sup>85</sup>, Gillian Mahy<sup>86</sup>, John Mansfield<sup>87</sup>, Dunecan Massey<sup>81</sup>, Christopher G. Mathew<sup>88,89</sup>, Dermot P. McGovern<sup>54</sup>, Raquel Milgrom<sup>90</sup>, Mitja Mitrovic<sup>91,55</sup>, , Craig Mowat<sup>92</sup>, William Newman<sup>77,78</sup>, Aylwin Ng<sup>93,7</sup>, Siew C. Ng<sup>94</sup>, Sok Meng Evelyn Ng<sup>66</sup>, Susanna Nikolaus<sup>23</sup>, Kaida Ning<sup>66</sup>, Markus Nöthen<sup>95</sup>, Ioannis Oikonomou<sup>66</sup>, Orazio Palmieri<sup>13</sup>, Miles Parkes<sup>81</sup>, Anne Phillips<sup>92</sup>, Cyriel Y. Ponsioen<sup>96</sup>, Uros Potocnik<sup>91,97</sup>, Natalie J. Prescott<sup>88,89</sup>, Deborah D. Proctor<sup>98</sup>, Graham Radford-Smith<sup>35,99</sup>, Jean-Francois Rahier<sup>100</sup>, Soumya Raychaudhuri<sup>69</sup>, Miguel Regueiro<sup>47</sup>, Florian Rieder<sup>2</sup>, John D. Rioux<sup>26,16</sup>, Stephan Ripke<sup>36,37</sup>, Rebecca Roberts<sup>21</sup>, Richard K. Russell<sup>67</sup>, Jeremy D. Sanderson<sup>101</sup>, Miquel Sans<sup>102</sup>, Jack Satsangi<sup>76</sup>, Eric E. Schadt<sup>34</sup>, Stefan Schreiber<sup>19,23</sup>, Philip L. Schumm<sup>103</sup>, Regan Scott<sup>47</sup>, Mark Seielstad<sup>104,105</sup>, Yashoda Sharma<sup>66</sup>, Mark S. Silverberg<sup>90</sup>, Lisa A. Simms<sup>35</sup>, Sarah L. Spain<sup>89</sup>, Hillary A. Steinhart<sup>90</sup>, Joanne M. Stempak<sup>90</sup>, Laura Stronati<sup>106</sup>, Jurgita Sventoraityte<sup>80</sup>, Stephan R. Targan<sup>54</sup>, Kristin M. Taylor<sup>101</sup>, Anje ter Velde<sup>96</sup>, Emilie Theatre<sup>57,58</sup>, Leif Torkvist<sup>107</sup>, Mark Tremelling<sup>108</sup>, Andrea van der Meulen<sup>109</sup>, Suzanne van Sommeren<sup>53</sup>, Eric Vasiliauskas<sup>54</sup>, Severine Vermeire<sup>110,33</sup>, Hein W. Verspaget<sup>109</sup>, Thomas Walters<sup>60,111</sup>, Kai Wang<sup>18</sup>, Ming-His Wang<sup>22,112</sup>, Rinse K. Weersma<sup>53</sup>, Zhi Wei<sup>113</sup>, , Cisca

Wijmenga<sup>55</sup>, David C. Wilson<sup>67,68</sup>, Juliane Winkelmann<sup>114,115</sup>, Ramnik J. Xavier<sup>7,37</sup>, Sebastian Zeissig<sup>23</sup>, Bin Zhang<sup>34</sup>, Clarence K. Zhang<sup>116</sup>, Hu Zhang<sup>117,118</sup>, Wei Zhang<sup>67</sup>, Hongyu Zhao<sup>116</sup>, Zhen Z. Zhao<sup>65</sup>, Australia and New Zealand IBDGC, Belgium IBD Genetics Consortium, Italian Group for IBD Genetic Consortium, NIDDK Inflammatory Bowel Disease Genetics Consortium, United Kingdom IBDGC, Wellcome Trust Case Control Consortium, Quebec IBD Genetics Consortium

<sup>1</sup>Department of Internal Medicine, Section of Digestive Diseases, Yale School of Medicine, New Haven, Connecticut, USA.

<sup>2</sup>Department of Gastroenterology and Hepatology, Digestive Disease Institute, Cleveland Clinic, Cleveland, Ohio, USA.

<sup>3</sup>Department of Pathobiology, Lerner Research Institute, Cleveland Clinic, Cleveland, Ohio, USA.

<sup>4</sup>Peninsula College of Medicine and Dentistry, Exeter, UK.

<sup>5</sup>Department of Gastroenterology, Erasmus Hospital, Brussels, Belgium, EU.

<sup>6</sup>Department of Gastroenterology, Free University of Brussels, Brussels, Belgium, EU.

<sup>7</sup>Gastroenterology Unit, Massachusetts General Hospital, Harvard Medical School, Boston, Massachusetts, USA.

<sup>8</sup>Division of Medical Sciences, Harvard Medical School, Boston, Massachusetts, USA.

<sup>9</sup>Medical Department, Viborg Regional Hospital, Viborg, Denmark, EU.

<sup>10</sup>Organ Center, Hospital of Southern Jutland Aabenraa, Aabenraa, Denmark, EU.

<sup>11</sup>Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus, Hinxton (Cambridge), UK.

<sup>12</sup>Inflammatory Bowel Disease Service, Department of Gastroenterology and Hepatology, Royal Adelaide Hospital, Adelaide, Australia.

<sup>13</sup>Unit of Gastroenterology, Istituto di Ricovero e Cura a Carattere Scientifico-Casa Sollievo della Sofferenza (IRCCS-CSS) Hospital, San Giovanni Rotondo, Italy.

<sup>14</sup>Unit of Gastroenterology SOD2, Azienda Ospedaliero Universitaria (AOU) Careggi, Florence, Italy.

<sup>15</sup>Department of Gastroenterology, Hôpital Maisonneuve-Rosemont, Montréal, Québec, Canada.

<sup>16</sup>Faculté de Médecine, Université de Montréal, Montréal, Québec, Canada.

<sup>17</sup>Division of Gastroenterology, Hepatology and Nutrition, Department of Medicine, University of Pittsburgh School of Medicine, Pittsburgh, Pennsylvania, USA.

<sup>18</sup>Center for Applied Genomics, The Children's Hospital of Philadelphia, Philadelphia, Pennsylvania, USA.

<sup>19</sup>Institute of Clinical Molecular Biology, Christian-Albrechts-University, Kiel, Germany.

- <sup>20</sup>Department of Gastroenterology and Hepatology, Flinders Medical Centre and School of Medicine, Flinders University, Adelaide, Australia.
- <sup>21</sup>Department of Medicine, University of Otago, Christchurch, New Zealand.
- <sup>22</sup>Meyerhoff Inflammatory Bowel Disease Center, Department of medicine, Johns Hopkins University School of Medicine, Baltimore, Maryland, USA.
- <sup>23</sup>Department for General Internal Medicine, Christian-Albrechts-University, Kiel, Germany.
- <sup>24</sup>Cardiovascular Health Research Unit, University of Washington, Seattle, Washington, USA.
- <sup>25</sup>Division of Gastroenterology, Royal Victoria Hospital, Montréal, Québec, Canada.
- <sup>26</sup>Research Center, Montreal Heart Institute, Montréal, Québec, Canada
- <sup>27</sup>Department of Medicine II, Ludwig-Maximilians-University Hospital Munich-Grosshadern, Munich, Germany.
- <sup>28</sup>Meyerhoff Inflammatory Bowel Disease Center, Department of Medicine, School of Medicine, and Department of Epidemiology, Bloomberg School of Public Health, Johns Hopkins University, Baltimore, Maryland, USA.
- <sup>29</sup>Department of Gastroenterology, Charit, Campus Mitte, Universitätsmedizin Berlin, Berlin, Germany.
- <sup>30</sup>IBD unit, Fremantle Hospital, Fremantle, Australia.
- <sup>31</sup>School of Medicine and Pharmacology, University of Western Australia, Fremantle, Australia.
- <sup>32</sup>Department of Genetics, Yale School of Medicine, New Haven, Connecticut, USA.
- <sup>33</sup>Department of Clinical and experimental medicine, Translational Research in GastroIntestinal Disorders (TARGID), Katholieke Universiteit (KU) Leuven, Leuven, Belgium.
- <sup>34</sup>Department of Genetics and Genomic Sciences, Mount Sinai School of Medicine, New York, New York, USA.
- <sup>35</sup>Inflammatory Bowel Diseases, Genetics and Computational Biology, Queensland Institute of Medical Research, Brisbane, Australia
- <sup>36</sup>Analytic and Translational Genetics Unit, Massachusetts General Hospital, Harvard Medical School, Boston, Massachusetts, USA.
- <sup>37</sup>Broad Institute of MIT and Harvard, Cambridge, Massachusetts, USA.
- <sup>38</sup>Department of Biosciences and Nutrition, Karolinska Institutet, Stockholm, Sweden.
- <sup>39</sup>IBD Center, Department of Gastroenterology, Istituto Clinico Humanitas, Milan, Italy.
- <sup>40</sup>Department of Gastroenterology and Hepatology, Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands.
- <sup>41</sup>Department of Hepatology and Gastroenterology, Ghent University Hospital, Ghent, Belgium.

- <sup>42</sup>Center of hepatology, Gastroenterology and Dietetics, Vilnius University, Vilnius, Lithuania.
- <sup>43</sup>Pediatric Gastroenterology, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio, USA.
- <sup>44</sup>Department of Gastroenterology, Université Catholique de Louvain (UCL) Cliniques Universitaires Saint-Luc, Brussels, Belgium.
- <sup>45</sup>Division of Gastroenterology, University Hospital Padua, Padua, Italy.
- <sup>46</sup>Department of Pediatrics, Cedars Sinai Medical Center, Los Angeles, California, USA
- <sup>47</sup>Division of Gastroenterology, Hepatology and Nutrition, Department of Medicine, University of Pittsburgh School of Medicine, Pittsburgh, Pennsylvania, USA
- <sup>48</sup>Department of Human Genetics, University of Pittsburgh Graduate School of Public Health, Pittsburgh, Pennsylvania, USA
- <sup>49</sup>Department of Gastroenterology, Torbay Hospital, Torbay, Devon, UK.
- <sup>50</sup>Center for Human Genetic Research, Massachusetts General Hospital, Harvard Medical School, Boston, Massachusetts, USA.
- <sup>51</sup>Pediatrics, Harvard Medical School, Boston, Massachusetts, USA.
- <sup>52</sup>Faculty of Medical & Health Sciences, School of Medical Sciences, The University of Auckland, Auckland, New Zealand.
- <sup>53</sup>Department of Gastroenterology and Hepatology, University Medical Center Groningen, Groningen, The Netherlands.
- <sup>54</sup>F.Widjaja Foundation Inflammatory Bowel and Immunobiology Research Institute, Cedars-Sinai Medical Center, Los Angeles, California, USA.
- <sup>55</sup>Department of Genetics, University Medical Center Groningen, Groningen, The Netherlands.
- <sup>56</sup>Department of Gastroenterology, Christchurch Hospital, Christchurch, New Zealand.
- <sup>57</sup>Unit of Animal Genomics, Groupe Interdisciplinaire de Génoprotéomique Appliquée (GIGA-R) Research Center, University of Liege, Liege, Belgium.
- <sup>58</sup>Faculty of Veterinary Medicine, University of Liege, Liege, Belgium.
- <sup>59</sup>Institute of Genetic Epidemiology, Helmholtz Zentrum München - German Research Center for Environmental Health, Neuherberg, Germany.
- <sup>60</sup>Gastroenterology, Hepatology and Nutrition, The Hospital for Sick Children, Toronto, Ontario, Canada.
- <sup>61</sup>Department of Pediatrics, University of Utah School of Medicine, Salt Lake City, Utah, USA.
- <sup>62</sup>Department of Gastroenterology, Faculty of Medicine and Health, Örebro University, Örebro SE- 70182, Sweden.
- <sup>63</sup>Department of Medicine, St Mark's Hospital, Harrow, Middlesex, UK.

- <sup>64</sup>Nottingham Digestive Diseases Centre, Queens Medical Centre, Nottingham, UK.
- <sup>65</sup>Genetic Epidemiology, Genetics and Computational Biology, Queensland Institute of Medical Research, Brisbane, Australia.
- <sup>66</sup>Section of Digestive Diseases, Department of Internal Medicine, Yale School of Medicine, NewHaven, Connecticut, USA.
- <sup>67</sup>Paediatric Gastroenterology and Nutrition, Royal Hospital for Sick Children, Edinburgh, UK.
- <sup>68</sup>Child Life and Health, University of Edinburgh, Edinburgh, Scotland, UK.
- <sup>69</sup>Division of Rheumatology Immunology and Allergy, Brigham and Women's Hospital, Boston, Massachusetts, USA.
- <sup>70</sup>Academy of Medicine, Lithuanian University of Health Sciences, Kaunas, Lithuania.
- <sup>71</sup>Wellcome Trust Centre for Human Genetics, University of Oxford, Headington, UK.
- <sup>72</sup>Christ Church, University of Oxford, St Aldates, UK.
- <sup>73</sup>Research Institute of Internal Medicine, Department of Transplantation Medicine, Division of Cancer, Surgery and Transplantation, Oslo University Hospital Rikshospitalet, Oslo, Norway.
- <sup>74</sup>Norwegian PSC Research Center, Department of Transplantation Medicine, Division of Cancer, Surgery and Transplantation, Oslo University Hospital Rikshospitalet, Oslo, Norway.
- <sup>75</sup>K.G. Jebsen Inflammation Research Centre, Institute of Clinical Medicine, University of Oslo, Oslo, Norway.
- <sup>76</sup>Gastrointestinal Unit, Wester General Hospital University of Edinburgh, Edinburgh, UK.
- <sup>77</sup>Genetic Medicine, Manchester Academic Health Science Centre, Manchester, UK.
- <sup>78</sup>The Manchester Centre for Genomic Medicine, University of Manchester, Manchester, UK.
- <sup>79</sup>Department of Pediatrics, Emory University School of Medicine, Atlanta, Georgia, USA.
- <sup>80</sup>Department of Gastroenterology, Lithuanian University of Medicine, Kaunas, Lithuania.
- <sup>81</sup>Inflammatory Bowel Disease Research Group, Addenbrooke's Hospital, Cambridge, UK.
- <sup>82</sup>Faculty of medicine, University of Latvia, Riga, Latvia.
- <sup>83</sup>Division of Pediatric Gastroenterology, Hepatology and Nutrition, Hospital for Sick Children, Toronto, Ontario, Canada.
- <sup>84</sup>Dipartimento di Neuroscienze, Psicologia, Area del Farmaco e Salute del Bambino (NEUROFARBA), Università di Firenze SOD Gastroenterologia e Nutrizione Ospedale pediatrico Meyer, Firenze, Italy.
- <sup>85</sup>Division of Gastroenterology, University Hospital CHU of Liege, Liege, Belgium.
- <sup>86</sup>Department of Gastroenterology, The Townsville Hospital, Townsville, Australia.
- <sup>87</sup>Institute of Human Genetics, Newcastle University, Newcastle upon Tyne, UK.
- <sup>88</sup>Department of Medical and Molecular Genetics, Guy's Hospital, London, UK.

- <sup>89</sup>Department of Medical and Molecular Genetics, King's College London School of Medicine, Guy's Hospital, London, UK.
- <sup>90</sup>Inflammatory Bowel Disease Centre, Mount Sinai Hospital, Toronto, Ontario, Canada.
- <sup>91</sup>Center for Human Molecular Genetics and Pharmacogenomics, Faculty of Medicine, University of Maribor, Maribor, Slovenia.
- <sup>92</sup>Department of Medicine, Ninewells Hospital and Medical School, Dundee, UK.
- <sup>93</sup>Center for Computational and Integrative Biology, Massachusetts General Hospital, Harvard Medical School, Boston, Massachusetts, USA.
- <sup>94</sup>Department of Medicine and Therapeutics, Institute of Digestive Disease, Chinese University of Hong Kong, Hong Kong.
- <sup>95</sup>Department of Genomics Life & Brain Center, University Hospital Bonn, Bonn, Germany.
- <sup>96</sup>Department of Gastroenterology, Academic Medical Center, Amsterdam, The Netherlands.
- <sup>97</sup>Faculty for Chemistry and Chemical Engineering, University of Maribor, Maribor, Slovenia.
- <sup>98</sup>Section of Digestive Diseases, Department of Medicine, Yale University, New Haven, Connecticut, USA.
- <sup>99</sup>Department of Gastroenterology, Royal Brisbane and Womens Hospital, Brisbane, Australia  
(3) School of Medicine, University of Queensland
- <sup>100</sup>Department of Gastroenterology, Université Catholique de Louvain (UCL) Centre hospitalier (CHU) Mont-Godinne, Mont-Godinne, Belgium.
- <sup>101</sup>Department of Gastroenterology, Guy's & St Thomas' NHS Foundation Trust, St-Thomas Hospital, London, UK.
- <sup>102</sup>Department of Digestive Diseases, Hospital Quiron Teknon, Barcelona, Spain.
- <sup>103</sup>Department of Public Health Sciences, University of Chicago, Chicago, Illinois, USA.
- <sup>104</sup>Human Genetics, Genome Institute of Singapore, Singapore.
- <sup>105</sup>Institute for Human Genetics, University of California San Francisco, San Francisco, California, USA.
- <sup>106</sup>Department of Biology of Radiations and Human Health, Agenzia nazionale per le nuove tecnologie l'energia e lo sviluppo economico sostenibile (ENEA), Rome, Italy.
- <sup>107</sup>Department of Clinical Science Intervention and Technology, Karolinska Institutet, Stockholm, Sweden.
- <sup>108</sup>Gastroenterology & General Medicine, Norfolk and Norwich University Hospital, Norwich, UK.
- <sup>109</sup>Department of Gastroenterology, Leiden University Medical Center, Leiden, The Netherlands.
- <sup>110</sup>Division of Gastroenterology, University Hospital Gasthuisberg, Leuven, Belgium.
- <sup>111</sup>Faculty of medicine, University of Toronto, Toronto, Ontario, Canada.

<sup>112</sup>Department of Gastroenterology and Hepatology, Digestive Disease Institute, Cleveland Clinic, Cleveland, Ohio, USA.

<sup>113</sup>Department of Computer Science, New Jersey Institute of Technology, Newark, New Jersey, USA.

<sup>114</sup>Institute of Human Genetics, Technische Universität München, Munich, Germany.

<sup>115</sup>Department of Neurology, Technische Universität München, Munich, Germany.

<sup>116</sup>Department of Biostatistics, School of Public Health, Yale University, NewHaven, Connecticut, USA.

<sup>117</sup>Department of Gastroenterology, West China Hospital, Chengdu, Sichuan, China.

<sup>118</sup>State Key Laboratory of Biotherapy, Sichuan University West China University of Medical Sciences (WCUMS), Chengdu, Sichuan, China.

## Members of the International PSC Study Group

Eva Ellinghaus<sup>1</sup>, Hugh Harley<sup>2</sup>, Peter Fickert<sup>3</sup>, Michael Trauner<sup>4</sup>, Emina Halilbasic<sup>4</sup>, Johan Fevery<sup>5</sup>, Werner Van Steenberghe<sup>6</sup>, Schalk Van der Merwe<sup>7</sup>, Chantal Housset<sup>8</sup>, Raoul Poupon<sup>8</sup>, Christophe Corpechot<sup>8</sup>, Verena Keitel<sup>9</sup>, Dieter Häussinger<sup>9</sup>, Ansgar Lohse<sup>10</sup>, Tim Lankisch<sup>11</sup>, Peter Sauer<sup>12</sup>, Peter Schirmacher<sup>13</sup>, Beate K. Straub<sup>13</sup>, Ruben Plentz<sup>14</sup>, Hugh E. Mulcahy<sup>15</sup>, Einar Björnsson<sup>16</sup>, Marco Marziani<sup>17</sup>, Ana Lleo<sup>18</sup>, Marco Carbone<sup>18</sup>, Carlo Selmi<sup>19</sup>, Luca Fabris<sup>20</sup>, Atsushi Tanaka<sup>21</sup>, Hiromasa Ohira<sup>22</sup>, Yoshiyuki Ueno<sup>23</sup>, Lars Aabakken<sup>24</sup>, Ewa Wunsch<sup>25</sup>, Andrzej Habior<sup>26</sup>, Alexander Knuth<sup>27</sup>, Beat Müllhaupt<sup>28</sup>, Andreas Geier<sup>29</sup>, Lina Lindstrøm<sup>30</sup>, Niklas Björkström<sup>30</sup>, Fredrik Rorsman<sup>31</sup>, Sven Almer<sup>32</sup>, Peter L. Jansen<sup>33</sup>, Andreas E. Kremer<sup>33</sup>, Frank G. Schaap<sup>34</sup>, Serge Zweers<sup>34</sup>, Pieter van der Vlies<sup>35</sup>, Cleo van Diemen<sup>35</sup>, Henk R. van Buuren<sup>36</sup>, David H. Adams<sup>37</sup>, Arthur Kaser<sup>38</sup>, Stephen Pereira<sup>39</sup>, George Webster<sup>40</sup>, Andrew Burroughs<sup>41</sup>, Shahid A Khan<sup>42</sup>, Simon D. Taylor-Robinson<sup>43</sup>, David Jones<sup>44</sup>, Alastair Burt<sup>45</sup>, Daniel Burger<sup>46</sup>, Kate Williamson<sup>47</sup>, David Shapiro<sup>48</sup>, Mario Strazzabosco<sup>49</sup>, Gregory Gores<sup>50</sup>, Keith Lindor<sup>51</sup>, Jayant Talwalkar<sup>52</sup>, David Goldberg<sup>52</sup>, Dennis Black<sup>53</sup>, Saul J. Karpen<sup>54</sup>, Christian Rust<sup>55</sup>

<sup>1</sup> Institute of Clinical Molecular Biology, Christian-Albrechts-University of Kiel, Kiel, Germany.

<sup>2</sup> University of Adelaide, Head of Clinical Hepatology, Co-Director of Viral Hepatitis Centre, Royal Adelaide Hospital, Australia.

<sup>3</sup> Division of Gastroenterology and Hepatology, Dept. of Medicine, Medical University of Graz, Graz, Austria.

<sup>4</sup> Department of Internal Medicine III, Division of Gastroenterology and Hepatology Medical University of Vienna Waehringer, Vienna, Austria.

<sup>5</sup> Laboratory of Hepatology, University Hospital Gasthuisberg, Leuven, Belgium.

<sup>6</sup> Department of Liver, Biliary and Pancreatic Diseases, University Hospital Gasthuisberg, Catholic University of Leuven, Leuven, Belgium.

<sup>7</sup> Laboratory of Hepatology, University Hospital Gasthuisberg, Leuven, Belgium.

<sup>8</sup> Service d'Hépatologie, Centre de référence des Maladies Inflammatoires des Voies Biliaires, Hôpital Saint-Antoine and UMR\_S938, Faculté de Médecine Saint-Antoine, Université Pierre et Marie Curie (UPMC), Paris, France.

<sup>9</sup> Department of Internal Medicine, Gastroenterology, Hepatology and Infectiology, Heinrich Heine Universität, Düsseldorf, Germany.

<sup>10</sup> I. Medizinische Klinik und Poliklinik, Universitätsklinikum Hamburg-Eppendorf, Hamburg, Germany.

- <sup>11</sup> Department of Gastroenterology Hepatology and Endocrinology, Hannover Medical School, Hannover, Germany.
- <sup>12</sup> Department of Internal Medicine IV, University Hospital of Heidelberg, Heidelberg, Germany.
- <sup>13</sup> Department of General Pathology, Institute of Pathology, Heidelberg, Germany.
- <sup>14</sup> Medizinische Universitätsklinik, Abteilung Innere Medizin 1, Tübingen, Germany.
- <sup>15</sup> Centre for Colorectal Diseases, St Vincent's University Hospital, Dublin, Ireland.
- <sup>16</sup> Department of Internal Medicine, Division of Gastroenterology and Hepatology, Landspítali University Hospital, Reykjavik, Iceland.
- <sup>17</sup> Department of Gastroenterology, Università Politecnica delle Marche, Ospedali Riuniti University Hospital, Ancona, Italy.
- <sup>18</sup> Center for Autoimmune Liver Diseases, Humanitas Clinical and Research Center, Rozzano, Milano, Italy.
- <sup>19</sup> Department of Translational Medicine, University of Milan, IRCCS Istituto Clinico Humanitas, Milan, Italy.
- <sup>20</sup> Department of Surgical and Gastroenterological Sciences, University of Padua, Padua, Italy.
- <sup>21</sup> Department of Medicine, Teikyo University School of Medicine, Tokyo, Japan.
- <sup>22</sup> Department of Gastroenterology and Rheumatology, Fukushima Medical University School of Medicine, Fukushima, Japan.
- <sup>23</sup> Division of Gastroenterology, Graduate School of Medicine, Tohoku University, Sendai, Japan.
- <sup>24</sup> Norwegian PSC Research Center, Division of Cancer, Surgery and Transplantation, Oslo University Hospital, Rikshospitalet, Oslo, Norway.
- <sup>25</sup> Liver Unit, Pomeranian Medical University, Szczecin, Poland.
- <sup>26</sup> Department of Gastroenterology and Hepatology, Medical Center for Postgraduate Education at the Maria Skłodowska-Curie Memorial Cancer Center, Institute of Oncology, Warsaw, Poland.
- <sup>27</sup> Clinic and Policlinic for Oncologie, University Hospital Zürich, Zürich, Switzerland.
- <sup>28</sup> Department of Hepatology and Gastroenterology, Swiss Hepato-Pancreatico-Biliary (HPB) and Transplant Center, University Hospital Zurich, Zurich, Switzerland.
- <sup>29</sup> Division of Gastroenterology and Hepatology, University Hospital Zürich, Zürich, Switzerland
- <sup>30</sup> Gastrocentrum Medicin, Karolinska Univ, Stockholm, Sweden.
- <sup>31</sup> Dept of Gastroenterology and Hepatology, Uppsala Univeristy Hospital, Uppsala, Sweden.
- <sup>32</sup> Avd. för Gastroenterologi och Hepatologi, Institutionen för Klinisk och Experimentell Medicin (IKE), The University Hospital, Linköping, Sweden.

- <sup>33</sup> Department of Gastroenterology & Hepatology, Tytgat Institute for Liver and Intestinal Research, Academic Medical Center, University of Amsterdam, The Netherlands.
- <sup>34</sup> Tytgat Institute for Liver and Intestinal research, Academic Medical Center, Amsterdam, The Netherlands.
- <sup>35</sup> Genome Analysis Facility, Dept. of Genetics, University Medical Centre Groningen, Groningen, The Netherlands.
- <sup>36</sup> Department of Gastroenterology and Hepatology, Erasmus University Medical Centre, Rotterdam, The Netherlands.
- <sup>37</sup> Centre for Liver Research, MRC Centre for Immune Regulation, Institute for Biomedical Research, University of Birmingham, Birmingham, B15 2TT, UK.
- <sup>38</sup> Section of Gastroenterology and Hepatology Medical Department, Addenbrooke's Hospital, Cambridge, UK.
- <sup>39</sup> The UCL Institute of Hepatology, Royal Free Hospital, London, UK.
- <sup>40</sup> Department of Gastroenterology, University College Hospital, London, UK.
- <sup>41</sup> Sheila Sherlock Liver Centre and Department of Surgery, Royal Free Hospital, London, UK.
- <sup>42</sup> Department of Gastroenterology, Hammersmith Hospital Campus, London, UK.
- <sup>43</sup> Department of Medicine, St Mary's Hospital Campus, Imperial College London, UK.
- <sup>44</sup> Institute of Cellular Medicine (Hepatology), The Medical School, Newcastle University, Newcastle upon Tyne, UK.
- <sup>45</sup> Clinical Deanery, Faculty of Medical Sciences, Newcastle University, Newcastle, UK.
- <sup>46</sup> Translational Gastroenterology Unit, John Radcliffe Hospital, Oxford Radcliffe NHS Trust, Oxford, UK.
- <sup>47</sup> Gastroenterology Department, John Radcliffe Hospital, Oxford University Hospitals, Oxford, UK.
- <sup>48</sup> 4350 La Jolla Village Drive Suite 960, San Diego, CA 92122, USA.
- <sup>49</sup> Department of Internal Medicine Section of Digestive Diseases Yale University, New Haven, CT, USA.
- <sup>50</sup> College of Medicine, Mayo Clinic, Rochester, MN, USA.
- <sup>51</sup> Division of Gastroenterology and Hepatology, Mayo Clinic, Rochester, MN, USA.
- <sup>52</sup> Division of gastroenterology, Dept. of Medicine, Hospital of the University of Pennsylvania and Perelman Center for Advanced Medicine, USA.
- <sup>53</sup> Memphis Medical Center, Memphis, Tennessee, USA.
- <sup>54</sup> Department of Pediatrics, Baylor College of Medicine, Houston, Texas, USA.
- <sup>55</sup> Department of Medicine 2, Grosshadern, University of Munich, Munich, Germany.