Supplementary Material from

Psoriasis is associated with increased beta-defensin genomic copy number

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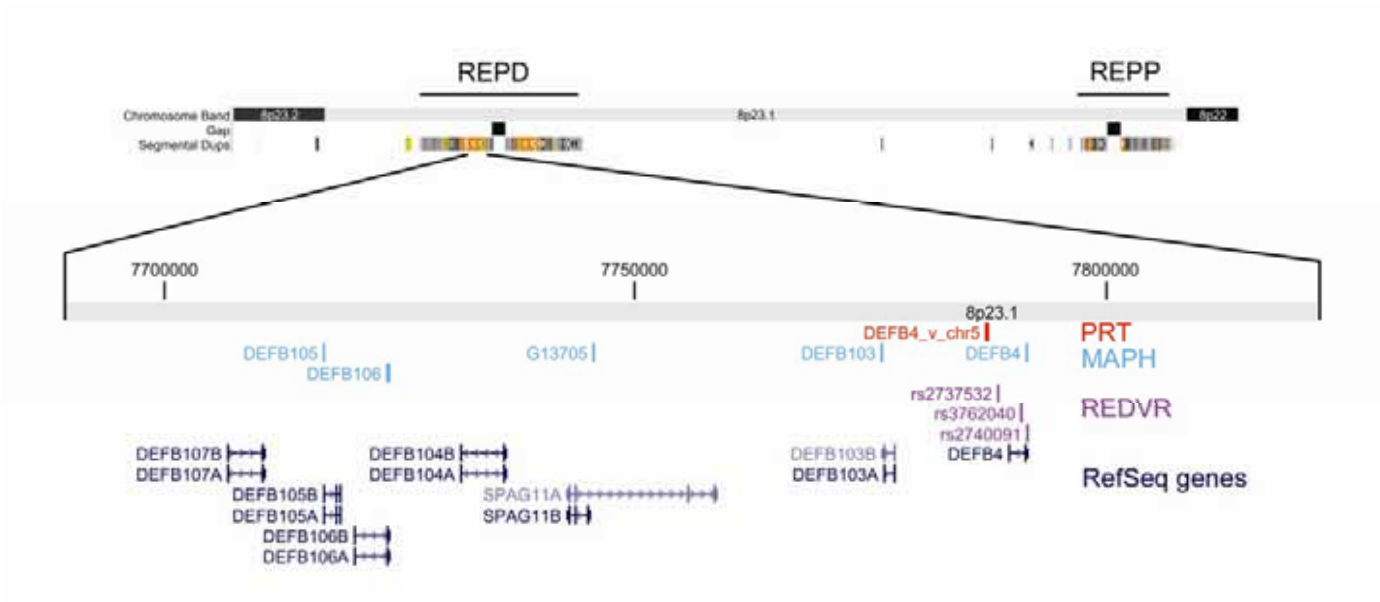
Supplementary Figure 1 Genomic structure of the copy-variable beta-defensin cluster on chromosome 8p23.1.

The chromosomal region is shown in the top half of the diagram, with assembly gaps, segmental duplications, and olfactory repeat regions REPD and REPP highlighted. The beta-defensin repeat region is within an olfactory repeat region, and an assembled example is shown expanded in the lower half of the diagram. This shows the region assayed in PRT, the variable nucleotides used for REDVR and the MAPH probes. Also shown are RefSeq genes, with both A and B versions shown. This nomenclature solely reflects the two beta-defensin repeats at REPD in this assembly, and hence their different physical locations, but not necessarily any sequence differences between the two coding regions of each gene. Figure based on UCSC Genome browser build 36.1.



Supplementary Methods for Hollox *et al*. "Psoriasis is associated with increased beta-defensin genomic copy number"

Section A: DNA samples

A1: Dutch samples (from Nijmegen)

Samples from 202 psoriatics were recruited from individuals referred to the out-patient clinic of the Department of Dermatology of the Radboud University Medical Centre by their general practitioner or dermatologist. Only patients diagnosed with psoriasis vulgaris were included. Patients with generalized pustular psoriasis or pustular psoriasis of hands and feet were excluded. The mean age of the psoriasis population was 54 (± 15) years, with a male:female ratio of 70:30. Examination of the medical records showed that 84% of the patients had been treated with UVB therapy, 76% had received at least one systemic therapy, and 18% were treated with biologicals (mainly anti-TNFα therapy). This classifies the Dutch patient group as having moderate to severe psoriasis. Control samples from the population of Nijmegen were obtained from the Nijmegen Biomedical Study (NBS)[1]. Additional control blood samples were from 64 healthy volunteers who were students or employees of the Radboud University Nijmegen Medical Centre. The mean age of the NBS controls was 54 (± 14) years with a male-female ratio of 70/30, to match that of the psoriasis patient group. The mean age of the combined control groups was 58 (± 13) years, with a male:female ratio of 56:44.

All controls and patients were of native European Dutch origin. Blood was stored at -80˚C and genomic DNA was isolated by standard procedures (either Qiagen column or salting-out procedures); DNA from all cases and about half the controls were prepared by the same (Qiagen) method. Permission for these studies was obtained from the local medical ethics committee (Commissie Mensgebonden Onderzoek).

A2: German samples (from Münster)

The German psoriasis cohort has been described previously[2]. In brief, 375 unrelated patients of German origin with psoriasis vulgaris were recruited through dermatology clinics at two psoriasis rehabilitation hospitals. An early onset form of psoriasis - mainly plaque type of psoriasis vulgaris - was diagnosed in all but three patients with an average age of onset of 23 ± 11 years. The mean age at the time of recruitment was 50 ± 12 years. 62 % of patients were male. All DNA samples were prepared in the same laboratory by the same methods. The study including recruitment of controls was approved by the ethical committee of the University of Münster. The 376 controls had no psoriasis vulgaris at the time of recruitment, when the average

age was 32 ± 10 years. All of them were healthy blood donors recruited in Northern Germany and 59 % were male. The investigations were conducted according to Declaration of Helsinki principles. All individuals gave their written informed consent.

Section B: Typing details and strategies

Current technologies for large-scale copy number determination are error-prone and require careful evaluation and quality control for use in robust case-control association studies. Our work on the Dutch cohort used two typing methods to provide quality-control comparisons. The Dutch samples were first typed for *DEFB4* copy number using MAPH supplemented by REDVR assays, and subsequently by PRT. Once PRT methods had been sufficiently developed and validated for stand-alone application, the German samples were typed by PRT only. The numbers of cases and controls successfully typed by each method were as follows:

	Total available	MAPH/REDVR (tests attempted)	PRT results (tests attempted)
Dutch cases	202	190 (202)	179 (179)
Dutch controls	317	303 (317)	272 (274)
German cases	375	-	319 (375)
German controls	376	-	305 (376)
Total	1270	493 (519)	1075 (1204)

B1: PRT methods

The details of paralogue ratio test (PRT) typing for the *DEFB4* copy-variable unit have already been published elsewhere[3]. Briefly, the PRT assay exploits a low-copy dispersed repeat (a processed heat-shock protein pseudogene) about 2kb upstream of *DEFB4*, and uses a single pair of precisely designed primers (HSPD5.8F CCAGATGAGACCAGTGTCC and FAM- or HEX-labelled primer HSPD5.8R TTTTAAGTTCAGCAATTACAGC) to amplify two products, one from the variable copy near *DEFB4* and one from an unlinked copy on chromosome 5 (which does not vary in copy number). Test and reference amplicons are distinguished by restriction digestion (*Hae*III) and electrophoresis. The primers are designed to have multiple mismatches to the other copies of this pseudogene in the genome[3]; in general, using a single primer pair to amplify both test and reference amplicons appears to allow greater precision in copy number measurements based on comparative PCR[3,4].

For each sample, two parallel PCRs were set up each containing 5-10ng of DNA and either HEX- or FAM-labelled primer. After PCR, the HEX- and FAM-labelled

products were mixed and digested with *Hae*III before separation by electrophoresis. The chr8(*DEFB4*):chr5 ratio was linearly related to the *DEFB4* copy number, and each experiment was calibrated either against copy numbers determined by MAPH/REDVR (for Dutch samples, see below), or on a standard set of 10 independent reference samples (for German samples).

As a quality filter, results were only accepted for use in copy number determination if the ratios of HEX- and FAM-labelled products differed by less than 15% of their mean. If accepted, the average of the two ratios was used in determining copy number. Further reassurance on the accuracy of copy number determination can be obtained from comparisons of the same samples with different typing methods and by clustering of results around integer values (see section C below).

In the PRT assay the Dutch samples were mostly typed in 96-well plate format including a mix of cases, controls and other samples in most experiments. Details of Dutch samples typed by PRT in different plates, together with summaries of residual analysis (see Section C), are shown in the table below. In mixed plates, cases and controls were allocated to random well positions so that cases and controls were interspersed on the same plate. All German samples were typed by PRT in batches of 72 samples on 96-well plates with 10 independent reference controls, each in duplicate. Apart from a single mixed plate, German psoriasis cases (4 plates) and controls (4 plates) were typed on separate plates, but summary statistics including residual analysis do not indicate any major differences between them (See section C below). Details of the experiment in which each sample was typed are included in the full listing of experimental data provided in **Supplementary Table 2** (separate document).

Summary of plate composition for PRT testing of Dutch samples, showing the numbers of cases and controls typed in each batch. For “mean residuals”, see section C. Results from batches M, N and R were part of batches of at least 20 samples including controls and/or samples from other studies. There are 339 PRT results from 272 control samples successfully typed, and 220 results from 179 cases; 95 samples were typed twice by PRT and 13 three times.

		plate/batch																
		A	B	C	D	E	F	G	H	J	K	L	M	N	P	Q	R	total
results	controls	42	28	52	32	24	26	29	23	38	11	8	1	1	11	12	1	339
obtained	cases	26	54	4	22	10	14	13	22	21	3	10	3	1	8	7	2	220
mean	signed	-0.0231	-0.0636	-0.0789	-0.0706	-0.0362	-0.0277	-0.0973	0.0262	-0.0118	-0.1486	-0.0760	0.0748	-0.1126	0.0003	-0.0350	0.0488	
residuals	absolute	0.1811	0.2025	0.2283	0.2023	0.2184	0.1848	0.2779	0.1987	0.2165	0.2127	0.2025	0.2282	0.1493	0.2537	0.1920	0.3764	

B2: MAPH methods

Details of MAPH methods for measurement of the beta-defensin cluster copy number have been published[5,6]. MAPH typing was carried out in a total of 12 experiments, in 9 of which cases and controls were typed together, and in 3 of which controls only were typed. All MAPH experiments were calibrated for copy number with three reference samples of known copy number, of which two reference samples were used as standards in all experiments. MAPH analysis in this study used an unweighted mean copy number estimate from the 5 probes used by Hollox et al.[6] (at *DEFB103*, *DEFB4*, *DEFB105*, *SPAG11* and *DEFB106*). Copy number values from each of these probes for each individual are shown in the full data listing of Supplementary Table 2. The sequences of all the probes used are listed in **Supplementary Table 1** (separate document).

B3: REDVR methods

To improve the accuracy of copy number measurement MAPH results were combined with REDVR (Restriction Enzyme Digest Variant Ratio) tests. REDVR methods, already successfully applied to the *DEFA1A3* copy number variation[7], indicate likely integer values for DNA samples based on proportions between variants (MultiSite Variants or MSVs) within the copy-variable repeat. Thus a sample with both forms of an MSV, in the ratio 1:1, suggests an even number of repeat units and thus is more likely to have a copy number of 4 than 5 if the MAPH measurement returns, for example, a value of 4.53.

Ratios of variants for three MSV positions, rs2740091, rs2737532 and rs3762040, were determined by PCR with fluorescently labelled primers and digestion with relevant restriction enzymes (details in table below). Quantification of signals allowed ratios of variants to be deduced, after minor correction for incomplete digestion (80-90%) presumably due to low-level heteroduplex formation – incomplete digestion was not observed in samples where only one form was present.

Variant	Primers	enzyme
rs2740091	5'-FAM-CAAACCAACATCACCCAATG AAATTGGCACCTGTGGTCTC	*Xmn*I
rs2737532	5'-FAM-GAAGAATGAGGTTGAAACCAGGA CAAAGCCACCATACCTCCAT	*Hpy*188I
rs3762040	5'-HEX-TCTCAAACTGCCCTTAGATCG ATGGGAAACTCTTATGAAGGAG	*Mbo*II

Among 493 samples with MAPH copy number measurements, 447 (91%) were informative for at least one of the variants tested, and 349 (71%) had all informative REDVR ratios consistent with the MAPH measurement rounded to the nearest integer. A further 53 (11%) gave REDVR ratios which appeared to rule out the nearest integer to the MAPH measurement, but suggested an

alternative integer consistent with values within the 95% confidence interval for MAPH copy number; for 20 samples (4%) different REDVR assays suggested different integers, one of which was consistent with the MAPH integer result. From comparisons with MLPA and PRT measurements on some of the same samples, we estimate the overall error rate for a single combined MAPH/REDVR copy number determination to be of the order of 5-10% per test (see section C4 below).

Section C: Quality control and accuracy

C1: Missingness and genotyping bias

In general, it has recently been established that spurious associations could be caused by differential bias in genotyping, even for well-established SNP typing methodologies; if the DNA preparation method for cases and controls differs sufficiently to cause unequal failure to score particular genotypes, or a genotyping bias applying differentially to cases or controls, this might be recorded as an apparent genotypic association[8]. This is especially relevant to copy number measurement, in which it is generally harder to produce satisfactory measurements of the higher copy numbers; differential drop-out of samples might therefore preferentially affect the highest copy numbers and thus lead to an apparent shift between cases and controls.

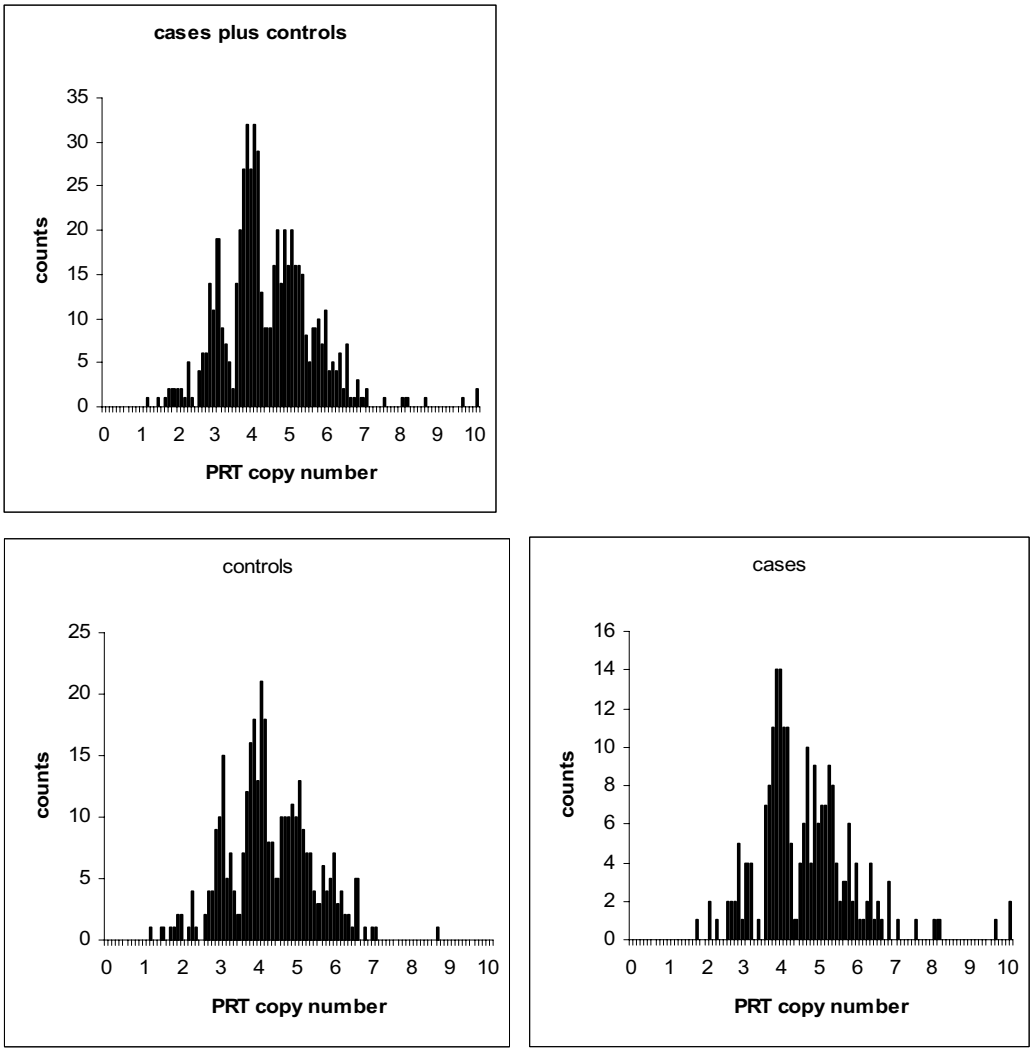
Methods of control and case DNA preparation were either closely matched for most samples (Dutch samples) or identical for all samples (German samples); see section A (“DNA samples”), above. The numbers of samples not producing a technically acceptable result (missingness) can be inferred from the summary table at the start of section B, above. The overall success rate of typing differs between the tests and between the cohorts, ranging from 81% (German PRT, controls) to 100% (Dutch PRT, cases). However, that difference is largely contributed by a sustained effort devoted to retyping failed Dutch samples by PRT, and in no data set is there a significant difference between the cases and controls in the number of failed tests (all P values >0.1).

In addition to the admixture of Dutch cases and controls on 96-well plates for PRT analysis, and the matched DNA preparation methods for the vast majority of samples, we can find no evidence that our approach has led to differential genotyping bias; comparisons of error as quantified by analysis of residuals (see below) fail to show any major differences between cases and controls sufficient to account for the association we observe.

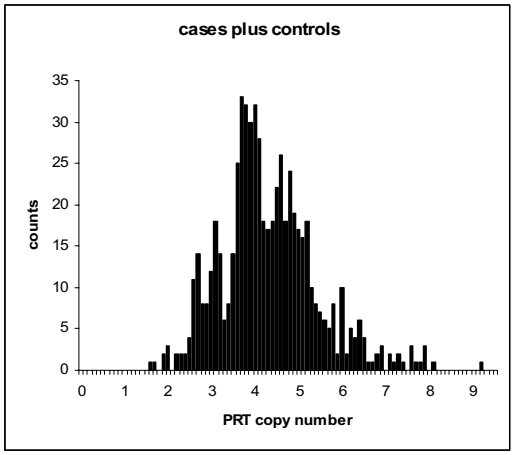
C2: Integer clustering and residuals

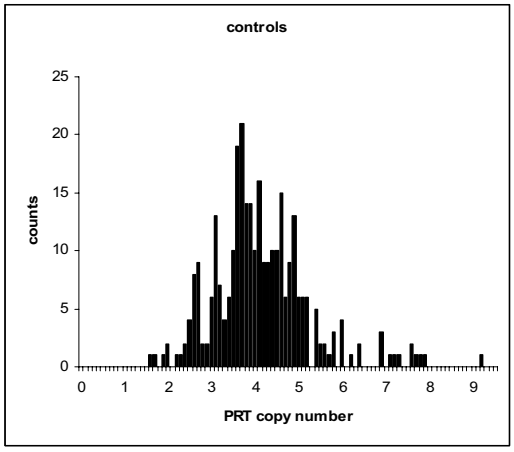
We assume that for analysis of germline predisposition to psoriasis or other phenotypes only integral copy numbers have biological meaning. Although mosaicism might in principle lead to the correct measurement of a non-integral copy number in a DNA sample, copy number measurements on the *DEFB4* cluster have failed to provide any evidence[3] that there are truly non-integral copy numbers (see also section C4, below). Our analysis therefore assumes that the differences between the measured value and the nearest integer (i.e., the residual values) result from measurement error and can thus be a useful index of measurement accuracy.

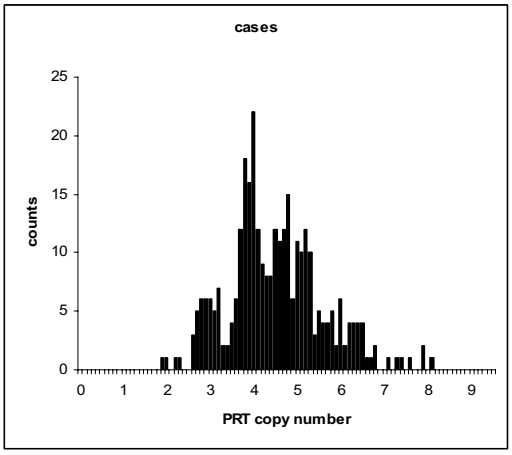
Any method based on ratios is predicted to determine lower copy number more accurately, and as expected PRT and MAPH data have clearer peaks around copy numbers of 2, 3 and 4 than around higher copy numbers:



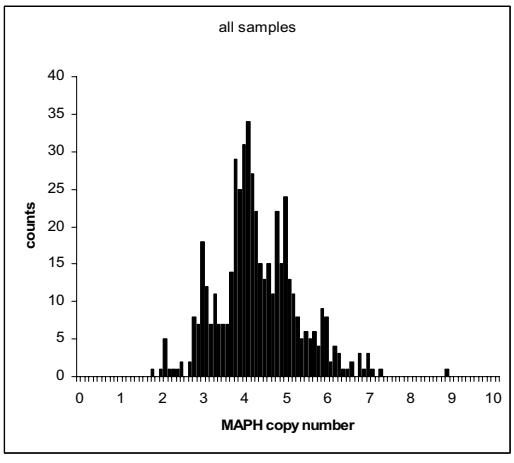
Clustering of PRT results around integer values for all Dutch samples (upper panel), and for controls (lower left) and cases (lower right) separately.

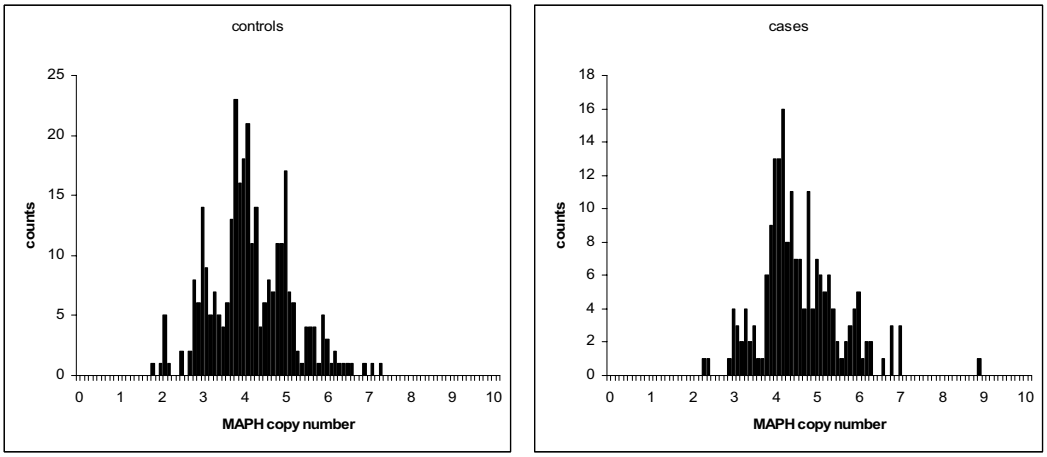






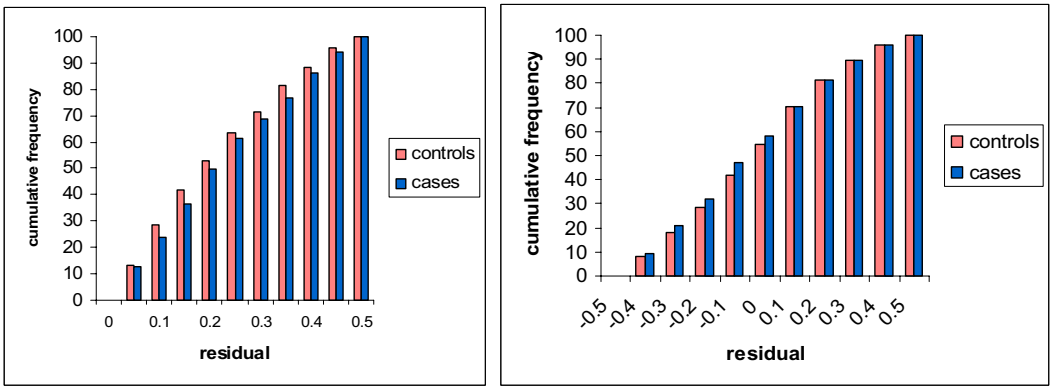
Clustering of PRT results around integer values for all German samples (upper panel), and for controls (lower left) and cases (lower right) separately.





Histograms of MAPH results for all Dutch samples (upper panel), and for controls (lower left) and cases (lower right) separately.

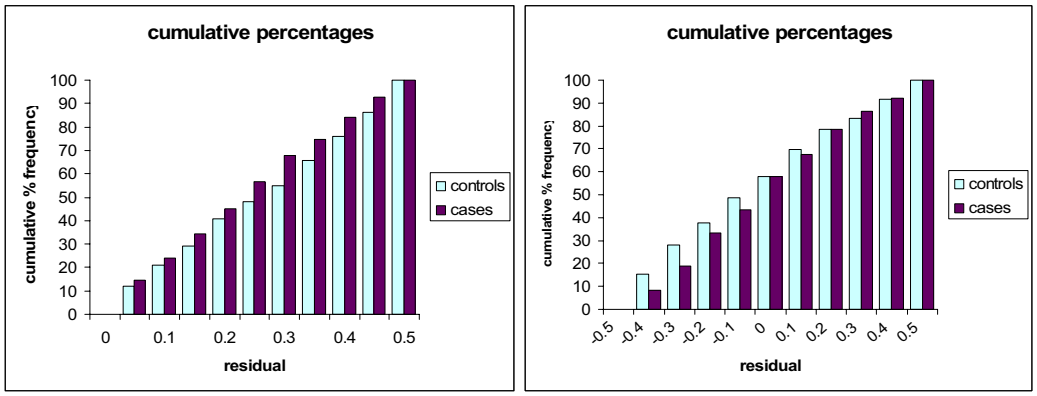
Cumulative frequencies of residual values are shown below, both as absolute values (i.e. magnitudes, ranging from 0 to 0.5) and as signed residuals, ranging from -0.5 to +0.5. In summary, absolute values of residuals are comparable between Dutch PRT cases (average residual value 0.22, standard deviation 0.14) and controls (0.21 ± 0.13) and between German PRT cases (0.23 ± 0.14) and controls (0.26 ± 0.15). Similarly, the signed values of residuals are also very similar between Dutch PRT cases (mean -0.055, SD 0.25) and controls (-0.038 ± 0.24) and between German PRT cases (-0.04 ± 0.27) and controls (-0.06 ± 0.29).



Cumulative frequency distributions of residuals from PRT data on Dutch controls and cases, displaying either absolute values (left panel) or signed values (right).

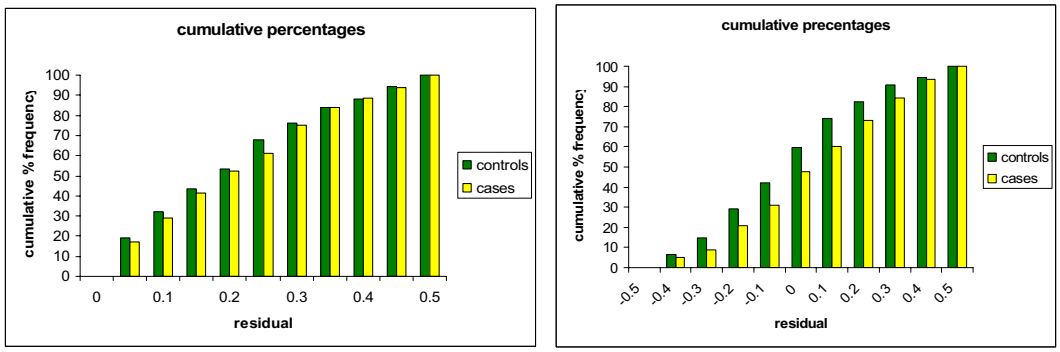
The distribution of signed residuals for PRT of German samples shows a small excess of larger negative residuals in controls relative to cases; however, although this difference is significant on analysis of absolute residual values ($P \approx 0.006$), the magnitude of the difference is small (an overall mean excess

residual of 0.03 repeat units per control sample), and the difference in signed residuals is not significant (P ≈ 0.13).



Cumulative frequency distributions of residuals from PRT data on German controls and cases, displaying either absolute values (left panel) or signed values (right).

Similarly, there is a small overall excess of negative residuals among Dutch MAPH controls (average magnitude 0.068 repeat units per sample) relative to cases that is statistically significant (P ≈ 0.0013), but is not evident in the distribution of absolute residuals (P ≈ 0.216):

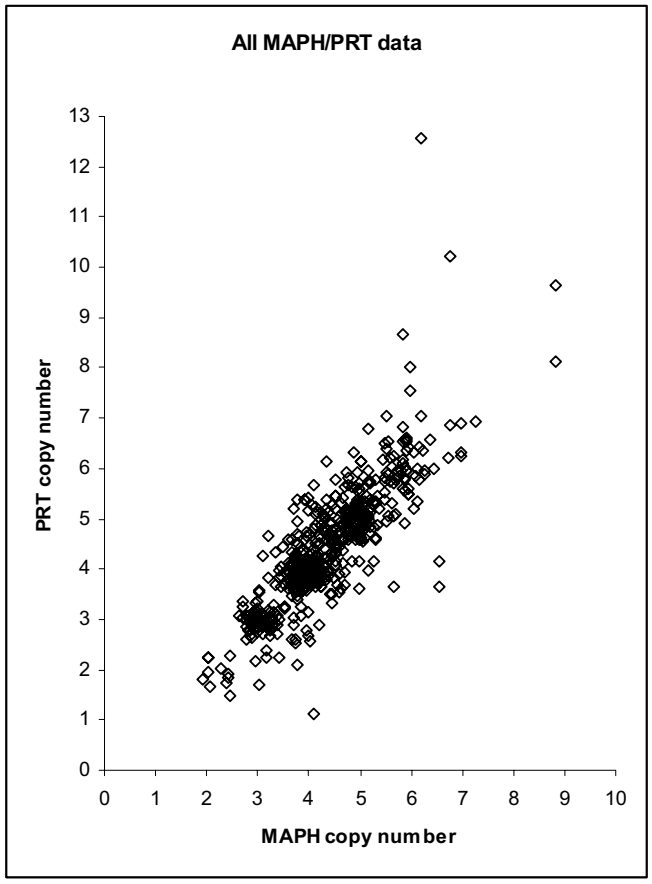


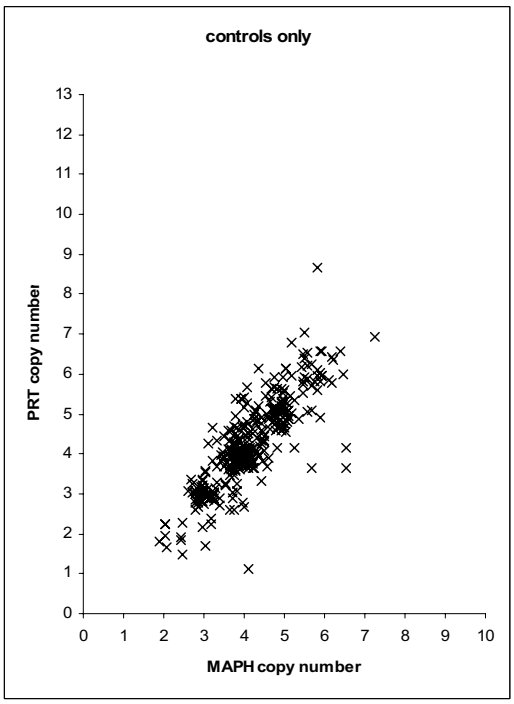
Cumulative frequency distributions of residuals from MAPH data on Dutch controls and cases, displaying either absolute values (left panel) or signed values (right).

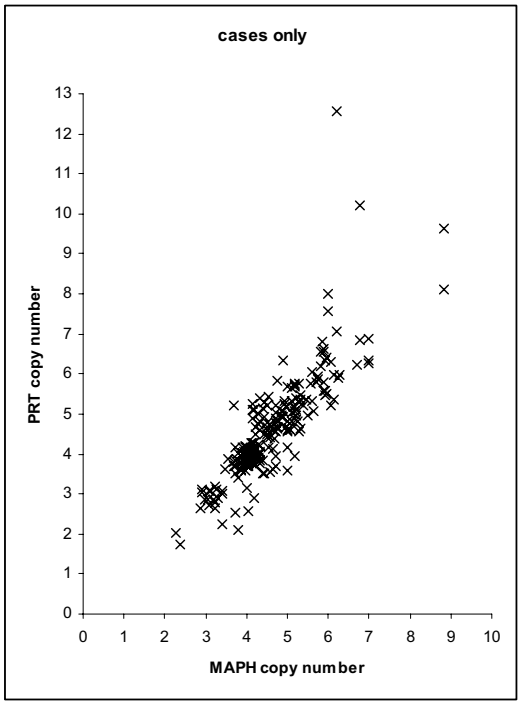
C3: Agreement between platforms

Reassurance about the accuracy of copy number determination can come from agreement between different typing platforms. Comparisons between PRT and MAPH data could be made for 451 Dutch samples; some of these had been typed more than once by PRT, either to investigate disagreement with MAPH results or to provide a measure of assay reproducibility. There were therefore a total of 559 pairwise comparisons that could be made between PRT and MAPH

data, of which 392 (70%) agreed on the integer-rounded result. Scatterplots of PRT and MAPH data for the same samples are shown:

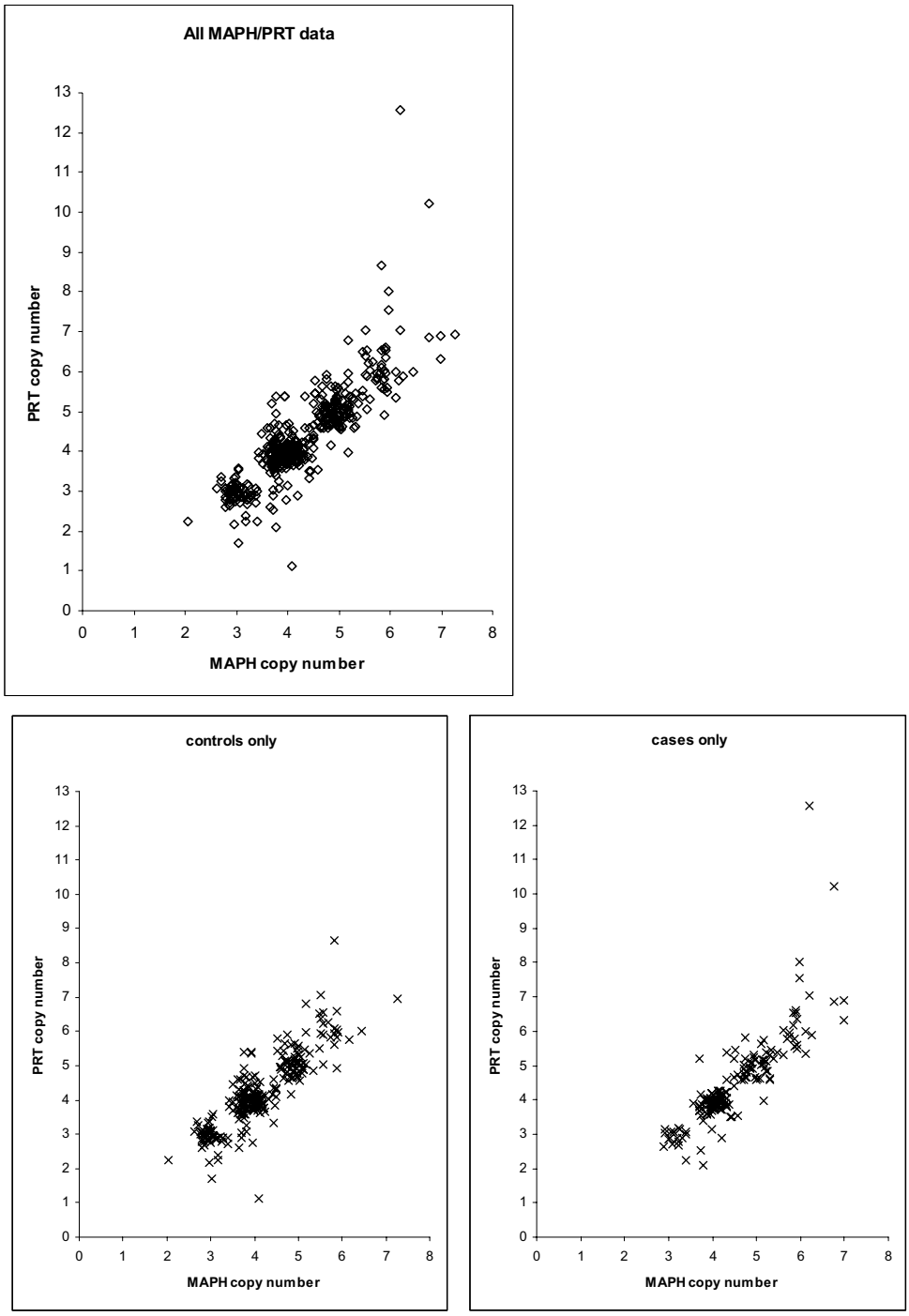






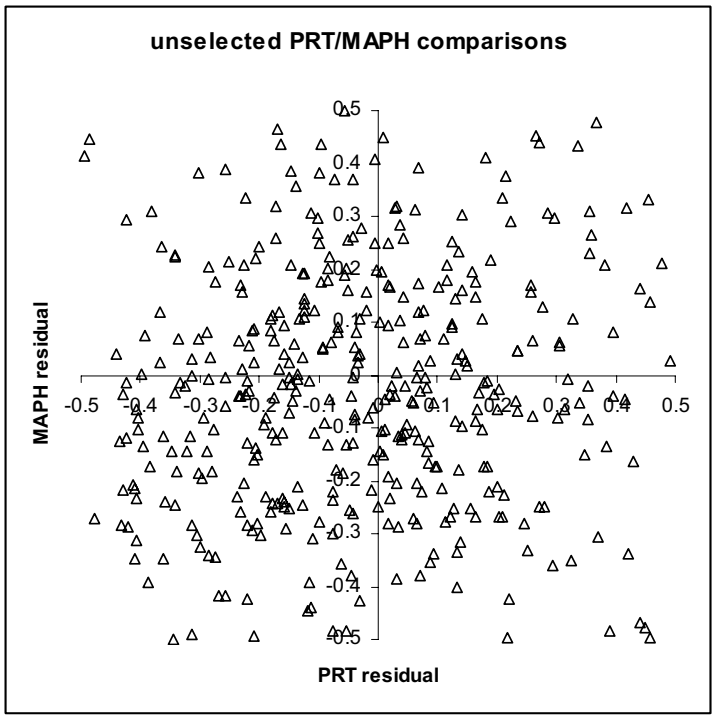
Scatterplots of PRT and MAPH data for all samples (top panel), and controls (left) and cases (right) shown separately.

The agreement between PRT and MAPH is significantly higher (336/415 ≈ 81%, $P \approx 1.6 \times 10^{-4}$) among the subset of samples for which REDVR data are consistent with the integer-rounded MAPH, suggesting that selecting MAPH data on the basis of consistency with REDVR data, or of agreement with PRT data, genuinely leads to increased accuracy of copy number determination. Scatterplots for these comparisons are shown below:



Scatterplots of PRT and REDVR-confirmed MAPH data for all samples (top panel), and controls (left) and cases (right) shown separately.

For the 392 pairwise comparisons in which PRT and MAPH agree on integral copy number, the residuals from PRT and from MAPH are not significantly correlated (R^2=0.0048) suggesting both that the true copy number is indeed an integer, and that there is no detectable systematic bias in copy number determination that is shared by both PRT and MAPH assays:



Comparison of integer residuals from the same 392 pairs of PRT and MAPH results.

C4: Error estimates

We have already published detailed analysis of the measurement error attributable to PRT and MAPH using comparisons between the data from samples typed by these methods and others, and the results of replicate testing[3]. Briefly, these studies on the Dutch samples concluded that incorrect calls of the integer copy number were strongly dependent on copy number, with lower copy numbers determined most accurately; the residual value was also clearly correlated with test error. We estimated an overall error rate for a single PRT of approximately 8% (95% confidence interval 5.6-11.8%).

Three-way comparisons between PRT, MAPH/REDVR and MLPA tests on a subset of 135 Dutch samples suggested that the frequency of incorrect integer calls for these three methods were approximately equal, and consistent with frequencies of about 5-10% per test[3]. Duplicate testing of 50 German samples by PRT indicated that although slightly higher than for the Dutch PRT measurements, the frequency of incorrect integer calls for a single PRT assay was similar, at approximately 11% (95% confidence interval 7%-17%).

Section D: Statistical Analysis

In all analyses a single null hypothesis was tested initially, that beta-defensin copy number and psoriasis status are independent. Although different approaches were taken in evaluating this hypothesis and the consequences of significant departure from it, a single genetic determinant (beta-defensin copy number) and a single phenotype (psoriasis) were evaluated, and so no corrections for multiple testing have been applied, as would be appropriate when several variants or several phenotypes has been analysed. For simplicity and consistency of presentation, the main manuscript shows the results of t-tests to compare the mean copy numbers of cases and controls.

To complement the statistical tests assuming no genotyping error, we carried out simulations to explore the effect of typing error on the significance of the difference between cases and controls. For the Dutch population the frequency of each copy number state was conservatively modelled by pooling cases and controls, and using the consensus values determined by comparison of MAPH, REDVR and PRT data (see Supplementary table 2). The program then used these copy number frequencies to simulate drawing a sample of 179 cases and 272 controls from the population, and the difference between the mean copy numbers of cases and control samples was determined. A run of 10^8 simulations was used to determine how often the difference exceeded that observed in the real data, to give the P values shown below. As expected, these simulations yield P-values similar to those derived from t-tests. This procedure was then repeated, but incorporating a realistic, copy-number dependent model of PRT error[3] applied to each individual in each simulation. These simulations were also run for the German data (319 cases, 305 controls), and for the Dutch and German samples combined:

Population	**P (no error)**	**P (with error function)**
Dutch	5.8×10^{-5}	1.4×10^{-4}
German	8.5×10^{-6}	2.0×10^{-5}
Combined data	3×10^{-8}(*)	3×10^{-8}

[* for the combined data without error, no simulations with a mean difference as high as that observed resulted from 100 million iterations, from which a conservative P value of 3×10^{-8} is derived]

These simulations assume that error rates are independent of the source of DNA, and apply equally to cases and controls. As a further safeguard, we also examined consensus copy numbers for the subset of Dutch DNA samples typed by PRT and known to have been prepared by the same extraction methods (121 controls and all 179 cases); copy numbers of these subsets of cases and controls considered on their own were also significantly different ($P= 2.9 \times 10^{-4}$

(t-test); simulation P values 2.2×10^{-4} without error, 4.4×10^{-4} including allowance for measurement error).

Among those Dutch samples for which clear age-of-onset data were available (n=133), there was no difference in mean age between individuals with 4 or fewer copies (29.4 years) versus individuals with more than 4 copies (28.5 years, p=0.72, t-test).

Simulation studies deriving λ_s

We performed simulation studies to determine what level of λ_s could be expected given the pattern of association we had observed. By linear regression of relative risk against copy number, the risk function λ_c=0.342c-0.512 was calculated, where c=copy number, and λ_c is the risk of psoriasis (relative to a population mean of 1) given the copy number c. Haplotype frequencies were inferred from the frequency of beta-defensin copy number per diploid genome (diplotype) by a maximum-likelihood approach [7,9]. Briefly, varying frequencies of copy number haplotype (frequencies f_0, f_1, f_2, f_3 and f_4 for copy number haplotypes of 0, 1, 2, 3 or 4) were iterated in steps of 0.01, and for each combination of frequencies, the exact joint probability of the full observed data set was calculated assuming Hardy-Weinberg equilibrium. This was calculated as $p_0 \times p_1 \times \ldots p_7$, where p_i is the probability of the observed number of individuals of copy number *i*. For example, the probability p_3 of observing exactly 54 people with 3 copies, given f_0 to f_3, is $[(2 \times f_0 \times f_3)+(2 \times f_1 \times f_2)]$[51]. The haplotype frequency distribution which maximised the total probability of the observed distribution in the Dutch controls was used (details in table below). We used the relative risk function and the beta-defensin copy haplotype frequencies of the control population to simulate populations of about 1.2×10^7 two-child families, of which approximately 2.4×10^5 families contained at least one child with psoriasis (representing a population frequency of 2%). Iteration of this simulation allowed us to estimate mean sibling recurrence risk (λ_s) attributable to beta-defensin copy number of 1.0754. Similar methods were also used to simulate affected sib pairs drawn from a population with the same haplotype frequencies and risk function for psoriasis, to estimate how frequently affected sib pair analyses would detect a significant linkage to the beta-defensin locus.

Copy number haplotype	**frequency**
1	0.17
2	0.59
3	0.22
4	0.02

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9. Aldred, P.M.R. Variation in human immune response genes. PhD Thesis, University of Nottingham (2004)

Supplementary table 1

MAPH probes designed for copy number analysis

Name	Type/region	Start coordinate (NCBI build 36.1)	End coordinate (NCBI build 36.1)	Reference
G13705	8p23 CNV	Chr8:7305559*	Chr8:7305765*	2
DEFB106	8p23 CNV	Chr8:7327364*	Chr8:7327683*	3
† DEFB104	8p23 CNV	Chr8:7315241*	Chr8:7315552*	3
DEFB103	8p23 CNV	Chr8:7275181*	Chr8:7275317*	2
DEFB105	8p23 CNV	Chr8:7334310*	Chr8:7334452*	3
DEFB4B	8p23 CNV	Chr8:7259889*	Chr8:7260004*	2
ST11G1	reference	Chr19:63775244	Chr19:63775469	1
PX3F11	X-linked	chrX:32547543	ChrX:32547885	5
nDLC1	reference	Chr8:13023765	Chr8:13023982	2
DEFB126	20p13	Chr20:74273	Chr20:74405	5
ST18A2	reference	Chr2:299006	Chr2:299246	1
ANGPT2	reference	Chr8:6347970	Chr8:6348154	2
DEFB125i	20p13	Chr20:25025	Chr20:25221	5
DEFB118	20q11	Chr20:29424518	Chr20:29424908	5
DEFB123	20q11	Chr20:29501452	Chr20:29501707	5
ST14A2	reference	Chr4:99218	Chr4:99638	1
ST18F2	reference	Chr7:158816019	Chr7:158816312	1
ST18C1	reference	Chr9:352611	Chr9:352841	1
DEFB122	20q11	Chr20:29472910	Chr20:29473174	5
DEFB124	20q11	Chr20:29516968	Chr20:29517163	5
DEFB119	20q11	Chr20:29428548	Chr20:29428988	5
DEFB114	6p12	Chr6:50035885	Chr6:50036185	5
SPLUNC2	reference	Chr20:31231963	Chr20:31232057	5
ST17A1	reference	Chr1:247074529	Chr1:247074685	1
AA01	reference	Chr8:8722885	Chr8:8723076	2
DEFB129	20p13	Chr20:158299	Chr20:158464	5
DEFB112	6p12	Chr6:50119215	Chr6:50119522	5
BPIL3	reference	Chr20:31090695	Chr20:31091167	5
LPLUNC1	reference	Chr20:31357427	Chr20:31357662	5
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LPLUNC4	reference	Chr20:31143822	Chr20:31144198	5
ST18G1	reference	Chr12:132169641	Chr12:132169740	1
DEFB120	20q11	Chr20:29440386	Chr20:29440815	5
MCM6	reference	Chr2:136314312	Chr2:136314839	5
LPLUNC3	reference	Chr20:31115023	Chr20:31115307	5
TBP	reference	Chr6:170706114	Chr6:170706362	4
GATA4	reference	Chr8:11653239	Chr8:11653641	2
ST19F2	reference	Chr5:180620496	Chr5:180620665	1
ST8D2	reference	Chr19:291949	Chr19:292411	1
ST19H1	reference	Chr15:100043567	Chr15:100043934	1
ST9A3	reference	Chr22:49488550	Chr22:49488897	1
SPLUNC1	reference	Chr20:31294502	Chr20:31295012	5
BPI	reference	Chr20:36373657	Chr20:36373761	5
ST7B2	reference	Chr16:18855*	Chr16:19224*	1
F11	reference	Chr4:187424329	Chr4:187424436	4
ST11G9	reference	Chr19:63772964	Chr19:63773090	1
LBP	reference	Chr20:36408360	Chr20:36408444	5
BPIL1	reference	Chr20:31070028	Chr20:31070365	5

n.b Defensin genes not described in the main paper but probes described here did not show evidence of full-length transcripts at the start of the project. None of these probes showed evidence of copy number variation.

† this probe was not used for copy number genotyping at 8p23.1
* full length of probe maps to two positions in this assembly

1. Hollox et al., 2002 J Med Genet. 39:790-5.
2. Hollox et al., 2003 Am J Hum Genet. 73:591-600
3. Hollox et al., 2005 J Negat Results Biomed. 4:9
4. Pickard et al. 2004 BMC Med Genet. 5:21
5. This paper

Supplementary data

Full sequence of all MAPH probes

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