

LD-CNV. Supplemental Figures and Tables

LD-CNV: rapid and simple discovery of chromosomal translocations using linkage disequilibrium between copy number variable loci

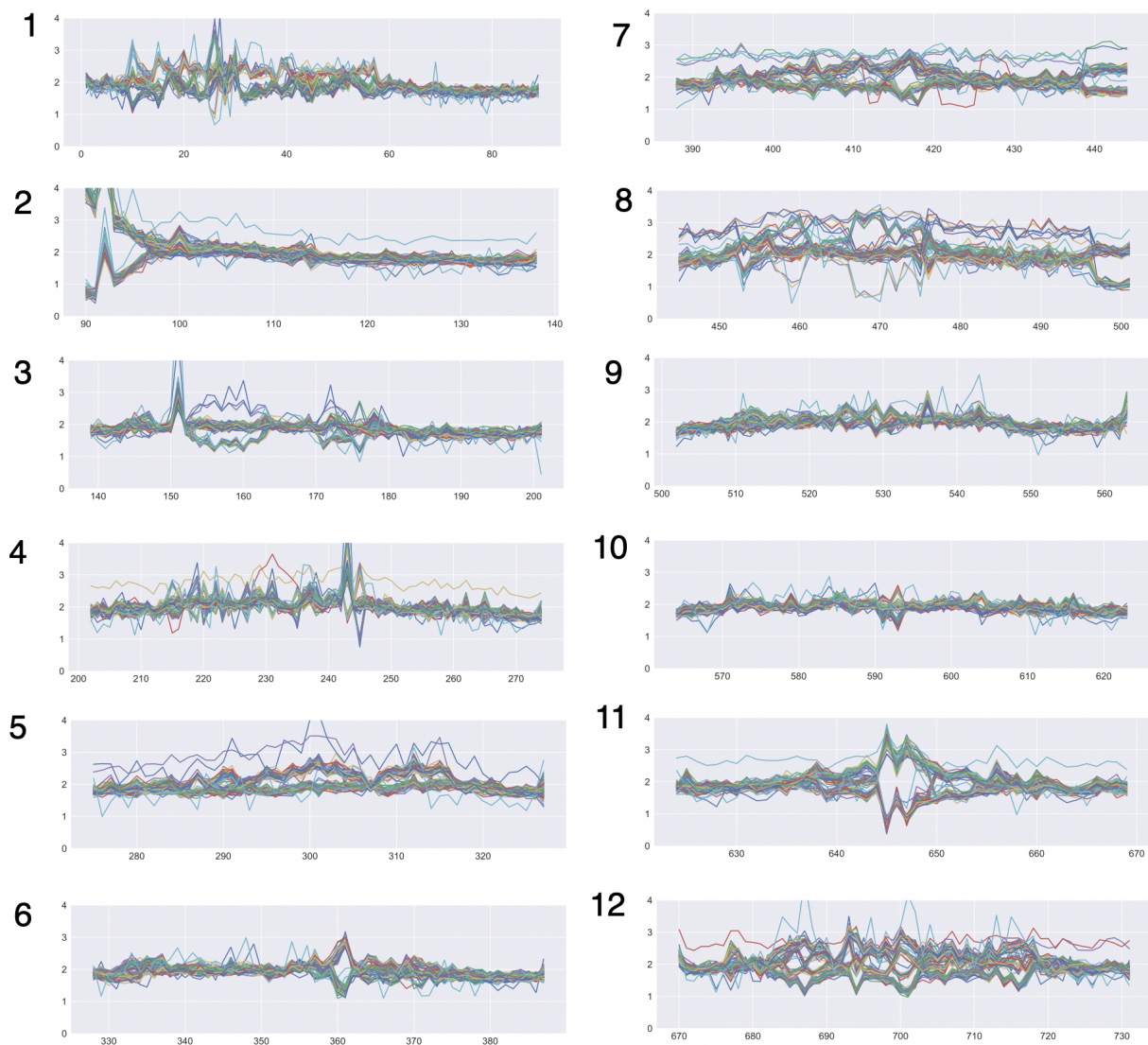
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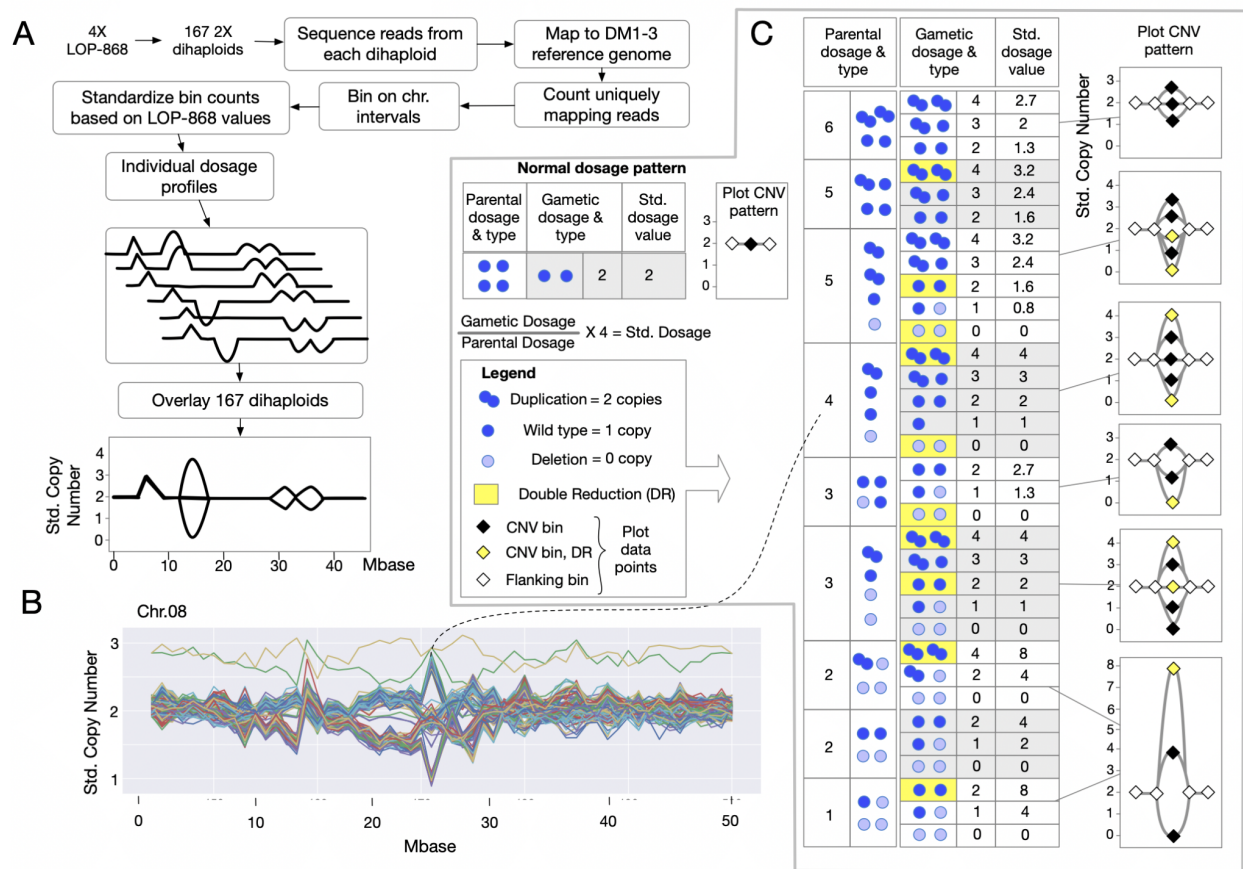
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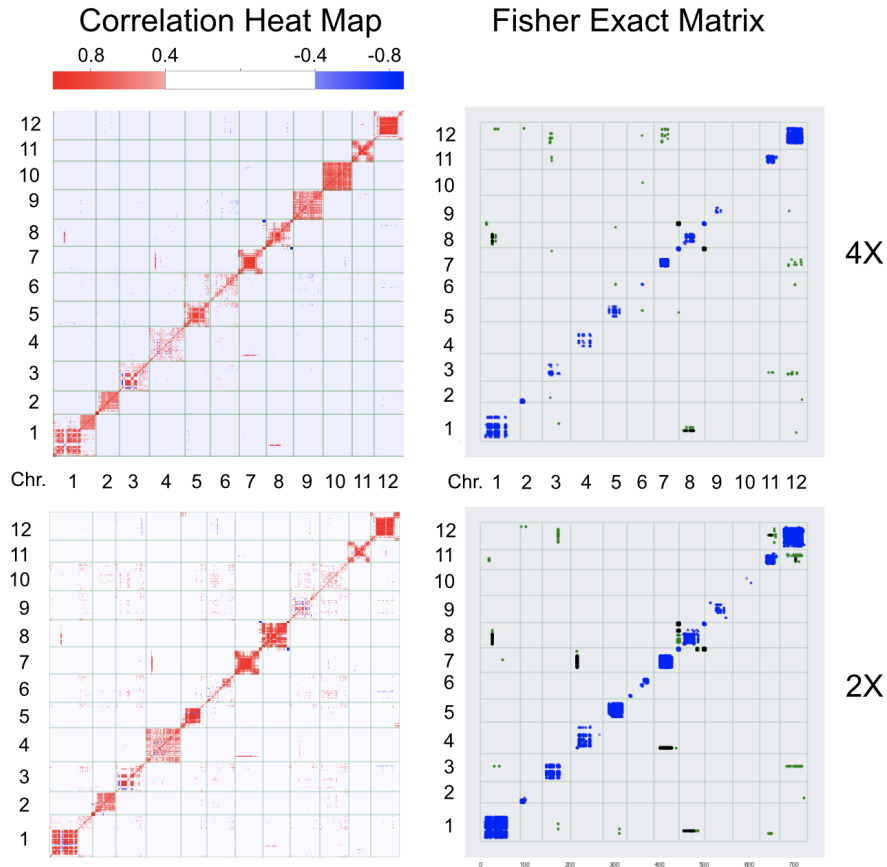
Supplemental Fig. 1. CNV patterns in BB, the dihaploid population of potato cv. PI 310467. The 12 chromosome dosage tracks of 84 2x individuals produced by crossing cv. PI 310467 to haploid inducer IvP48. The outlying tracks evident in some chromosomes, such as 7 and 8, represent trisomic individuals.



Supplemental Fig. 2. Analysis of CNV.

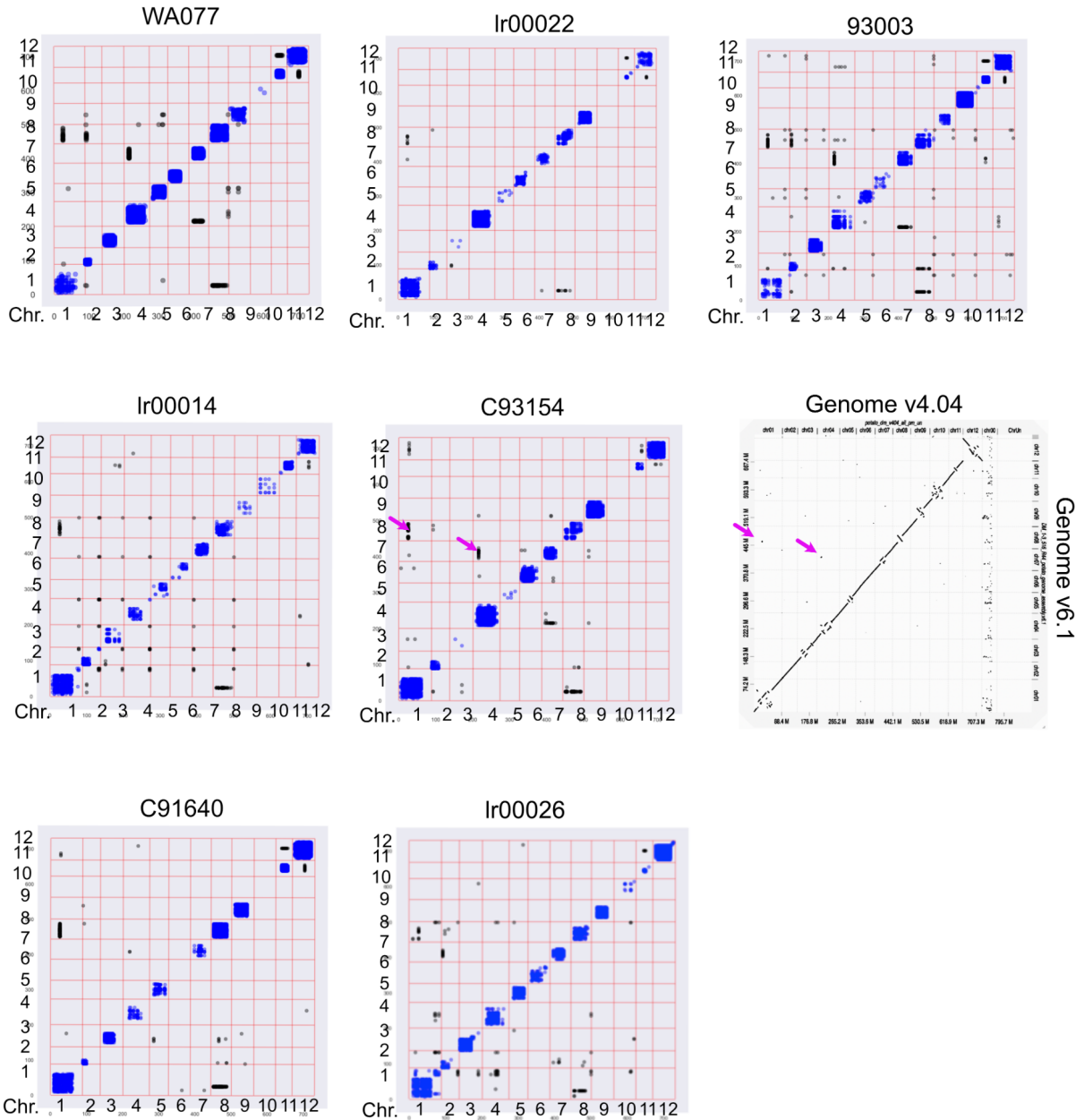
A. Overview of method to produce the dosage plot using a dihaploid family produced from an autotetraploid parent (LOP868 is the CIP accession name for Alca Tarma, the parent of the LOP population). B. Example of dosage plot resulting from the plotting of dihaploid profiles. Each color represents an individual. The two outliers are trisomics for chr. 8. C. Dosage variation in the autotetraploid parent and resulting segregation patterns in the dihaploids, which represent the maternal gametes. The “Std. Dosage Value” column represents the dihaploid dosage for a given locus relative to the dosage in the tetraploid parent. It is calculated from the formula: (Gametic dosage/parental dosage) * 4. It corresponds to the value displayed in B.

Dark blue: present allele. Light blue: absent allele (deletion). Overlapping circles represent a duplicated allele inherited as a unit. Yellow cells represent gametes formed by double reduction. Double reduction occurs at low frequency and is dependent on the recombination frequency between the locus of interest and the centromere.

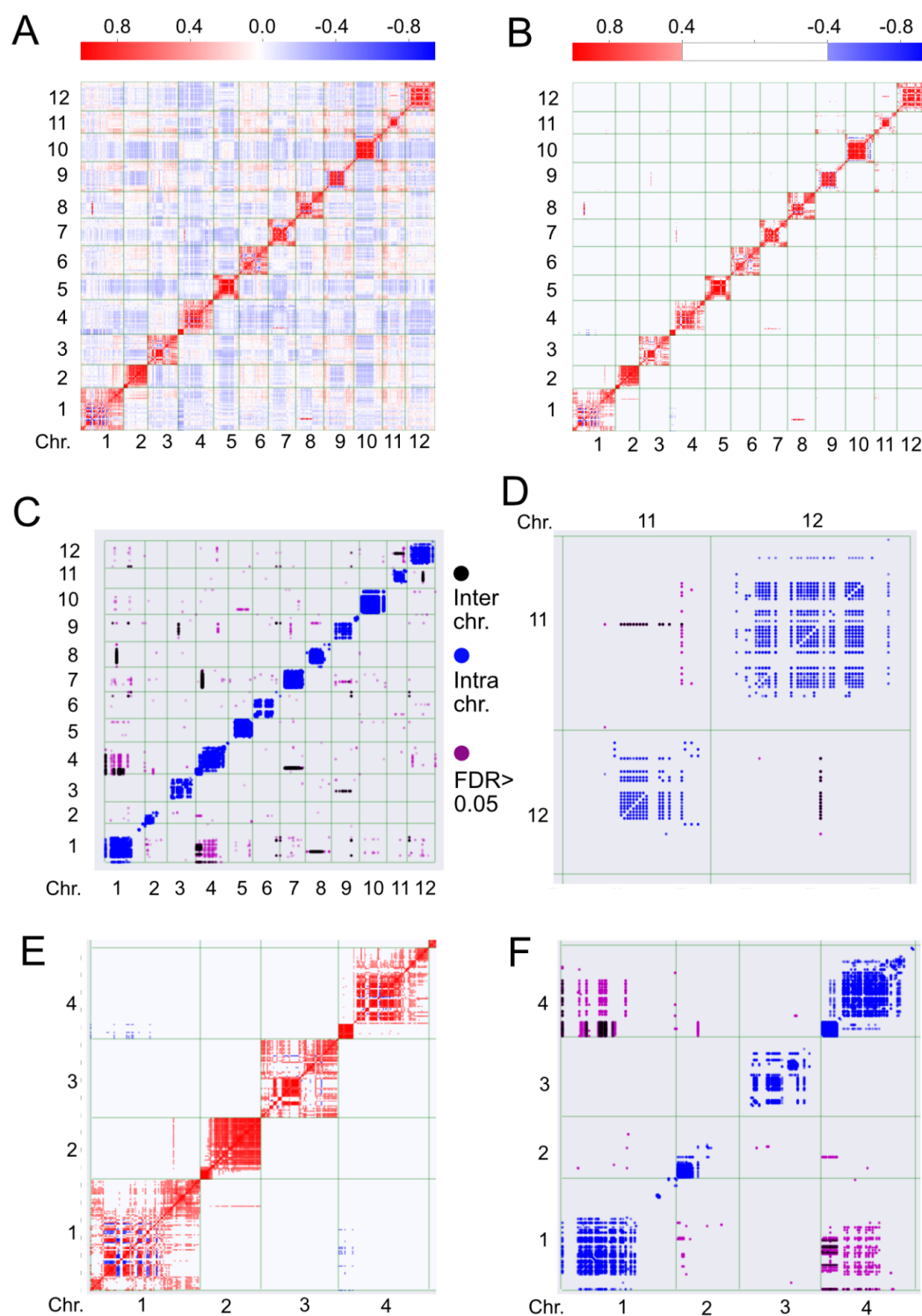


Supplemental Fig. 3. Effect of ploidy and statistical method on LD analysis.

4X analyses used the tetraploids of the BB population. 2X analyses used the diploids. The use of the Pearson's Correlation matrix and the implementation of the FET probability matrix are presented in Methods and are available at https://github.com/lcomai/cnv_mapping.

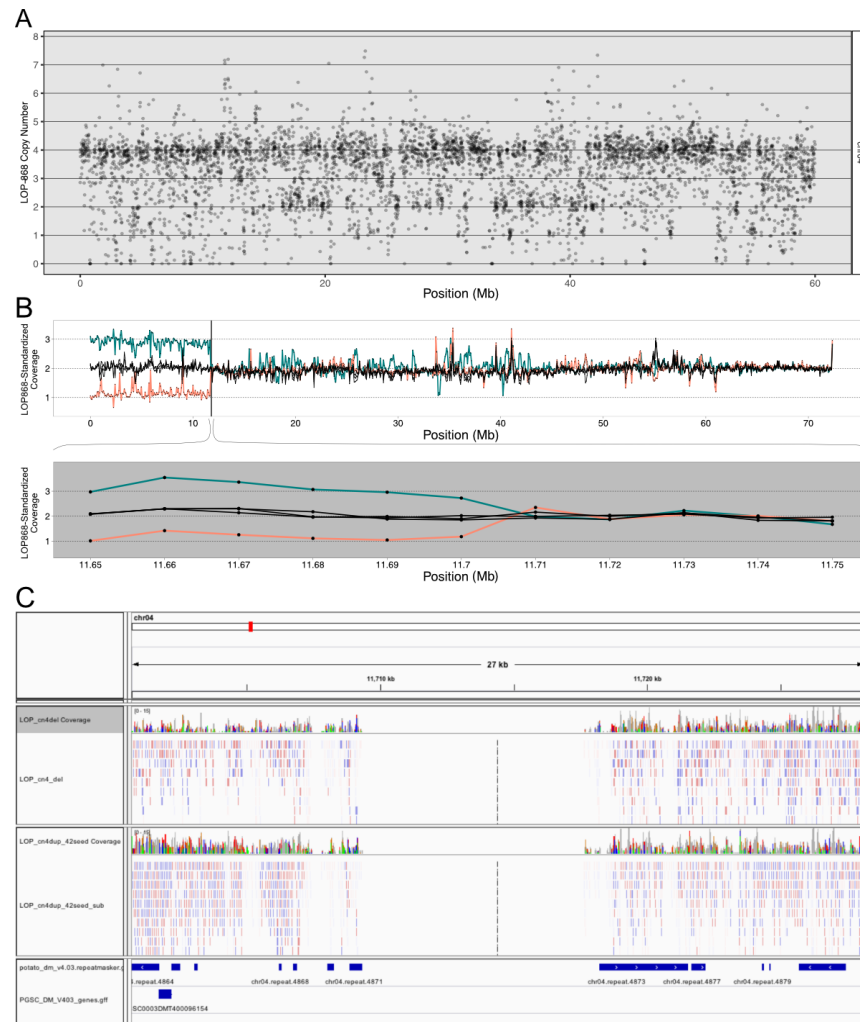


Supplemental Fig. 4. FET probability matrices from seven potato populations. The name of each dihaploid family seed parent is shown on top of each matrix. Blue: intrachromosomal LD. Black: interchromosomal LD. In all cases, the interchromosomal signals are consistent with pericentromeric translocation or misassembly during construction of the reference genome. The 8th matrix is a dot matrix comparison (Cabanettes and Klopp 2018) of the genome assembly version used in this work (4.04) to a more recent one (6.1). The magenta arrows point to regions in chromosome 8 vs 1 and 7 vs 4 that were misassembled in 4.04 and appear “translocated” in multiple population analyses. The absence of the signal in some populations could be due to the lack of segregating CNV in those regions.



Supplemental Fig. 5. Analysis of the LOP dihaploid family and comparison of Pearson's Correlation and Fisher's exact test for detection of correlated CNV sites.

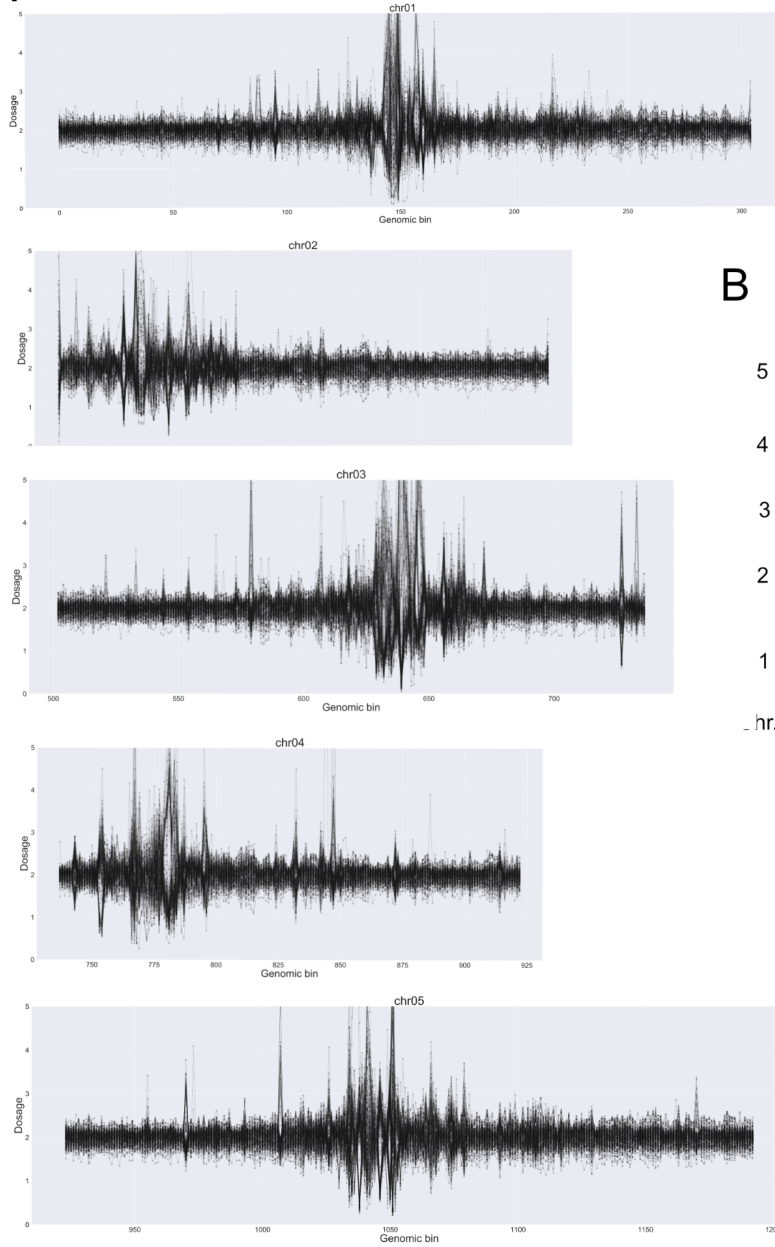
A. Raw correlation heatmap is less specific and more difficult to interpret than the FET probability matrix. B. Improved heatmap display by filtering low correlation signals. C. Clear demarcation of candidate correlated regions by cluster analysis and Fisher's exact test. A major signal is visible for chromosome 1 and 4. D. Fisher's exact test identifies candidate regions in chr. 11 and 12. This appears to be the same signal displayed by the BB population, suggesting that two different cultivated potatoes share polymorphism. E, F. Side by side comparison of translocation signals in filtered correlation and Fisher's Exact matrices.



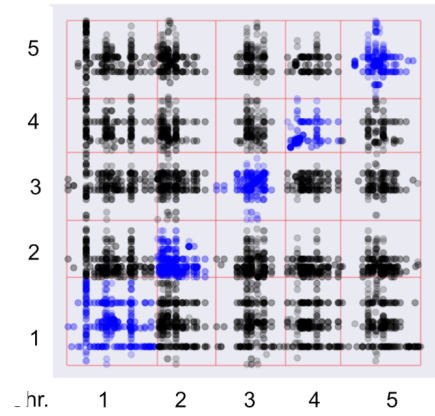
Supplemental Fig. 6. Breakpoint analysis of chromosome 4 short arm CNV.

A) Relative coverage in 4x Alca Tarma over contiguous non-overlapping 1Mb bins of chromosome 4. The affected euchromatic arm is dosage neutral (4 copies). B) Top track: relative mean read coverage in different pools of dihaploids derived from Alca Tarma binned in nonoverlapping 75kb bins. The lines are colored to represent duplicated (3 copies, teal, n=28 dihaploids) or deleted (pink, n=11 dihaploids) copy number states. Shown in black are three control groups, each composed of 20 randomly selected dihaploids with neither the duplicated nor deleted state. Bottom track: the zoomed-in gray region displays higher detail (10kb bins), illustrating the change from 3 or 1 to 2 copies between 11,700,000-11,710,000 bp. C) Browser view bracketing the chromosome 4 breakpoint. The top track displays reads of pooled one-copy dihaploids, the middle track the reads of pooled 3-copy dihaploids. The breakpoint is flanked by repeats 4871 and 4873, both of which are annotated as LTR/Gypsy retrotransposons.

A



B



Supplemental Fig. 7. Analysis of CNV in Arabidopsis natural accessions.

A. Chromosomal dosage profiles (chr. 1 to chr. 5, top to bottom) for the 192 *A. thaliana* accessions in Supplemental Table 1. B. FET probability matrix displaying the analysis of dosage state correlation for bins with FDR = 0.05 and for which the dosage difference between peaks is

at least 1. Black: interchromosomal, blue: intrachromosomal. The interchromosomal signals demonstrate LD common to most pericentromeric regions and therefore likely caused by repeats.

Supplemental Table 1. List of potato accessions and sequencing library deposit

Population Name	Tetraploid parent	Haploid inducer	Project number in NCBI read sequence database, Gb of data
LOP	Alca Tarma, a.k.a LOP 868	PL-4, IvP101	PRJNA408137, 313
BB	PI 310467, cv. Desiree in GRIN NRSP-6 germplasm collection	IvP48	PRJNA750855, estimated 300; Binned dosage values are at https://doi.org/10.25338/B88D2V
WA077	WA077 (CIP397077.16 or cv. Alliance/Sarnav)	PL-4, IvP101	PRJNA699631, 1053
lr00014	lr00014 (CIP300056.33)		
lr00022	lr00022 (CIP300072.1)		
lr00026	lr00026 (CIP300093.14)		
93003	93003 (CIP390637.1)		
C91640	C91640 (CIP388615.22)		
C93154	C93154 (CIP392820.1 or cv. BARI Alu-73)		

Supplemental Table 2. List of Arabidopsis accessions, SRA number and sequencing library characteristics

Accession + SRR (run name in SRA db at NCBI PRJNA273563)	Reads	Reads/MB
admixed-GMI-10_SRR1946059	59916068	500686.84
admixed-GMI-11_SRR1945728	24022752	200745.41
admixed-GMI-12_SRR1945997	29659042	247844.9
admixed-GMI-13_SRR1946061	33793413	282393.65
admixed-GMI-14_SRR1946077	34644790	289508.16
admixed-GMI-15_SRR1945983	19010462	158860.36
admixed-GMI-1_SRR1945626	19800036	165458.41
admixed-GMI-2_SRR1946071	28704624	239869.34
admixed-GMI-3_SRR1945491	40568849	339012.38
admixed-GMI-4_SRR1946022	36515150	305137.77
admixed-GMI-5_SRR1945490	16039451	134033.2
admixed-GMI-6_SRR1945623	19421444	162294.72
admixed-GMI-7_SRR1946069	21392755	178767.92
admixed-GMI-8_SRR1946070	30120943	251704.77
admixed-GMI-9_SRR1945638	32841499	274439.01
admixed-Monsanto-10_SRR1945537	9055319	75670.5
admixed-Monsanto-11_SRR1946183	16810601	140477.29
admixed-Monsanto-12_SRR1946126	17673420	147687.41
admixed-Monsanto-13_SRR1946184	21228594	177396.12
admixed-Monsanto-14_SRR1946471	13767340	115046.37
admixed-Monsanto-15_SRR1946400	12540265	104792.35
admixed-Monsanto-1_SRR1945940	10699758	89412.21
admixed-Monsanto-2_SRR1945519	15019671	125511.43
admixed-Monsanto-3_SRR1946108	17166146	143448.39

admixed-Monsanto-4_SRR1946149	15577834	130175.71
admixed-Monsanto-5_SRR1946418	5990357	50058.24
admixed-Monsanto-6_SRR1946135	17295878	144532.49
admixed-Monsanto-7_SRR1945556	9700463	81061.63
admixed-Monsanto-8_SRR1946479	17924451	149785.14
admixed-Monsanto-9_SRR1946438	13674684	114272.09
asia-GMI-1_SRR1946559	28507994	238226.21
asia-GMI-2_SRR1946560	55378823	462771.49
asia-GMI-3_SRR1946561	41916903	350277.36
asia-GMI-4_SRR1946558	25864327	216134.48
asia-GMI-5_SRR1946555	27246541	227684.91
asia-GMI-6_SRR1946566	35257170	294625.49
asia-GMI-7_SRR1946557	24278378	202881.54
asia-GMI-8_SRR1946565	41394961	345915.76
asia-GMI-9_SRR1946556	27748807	231882.08
asia-Monsanto-10_SRR1946049	30564177	255408.64
asia-Monsanto-11_SRR1946212	12655516	105755.44
asia-Monsanto-12_SRR1946225	12970324	108386.13
asia-Monsanto-13_SRR1946218	9342590	78071.08
asia-Monsanto-14_SRR1946312	9520464	79557.47
asia-Monsanto-15_SRR1946203	13293030	111082.81
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asia-Monsanto-2_SRR1946214	9013359	75319.87
asia-Monsanto-3_SRR1946219	8855134	73997.66
asia-Monsanto-4_SRR1946224	9790332	81812.62
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asia-Monsanto-7_SRR1946221	12692203	106062.02

asia-Monsanto-8_SRR1946050	23325425	194918.22
asia-Monsanto-9_SRR1946216	11455734	95729.5
central_europe-GMI-1_SRR1946008	19335113	161573.3
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central_europe-GMI-4_SRR1946562	63443436	530163.19
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western_europe-GMI-1_SRR1945657	26646916	222674.16
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western_europe-Monsanto-11_SRR1945553	7896785	65989.25
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