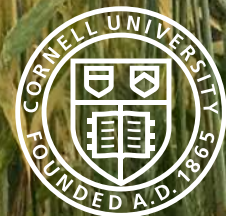


Challenges and opportunities in positional cloning and structural variation in polyploid crops

Ellie Taagen, PhD Candidate, Dr. Mark Sorrells Lab
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Plant and Animal Genome Conference, January 8th, 2022

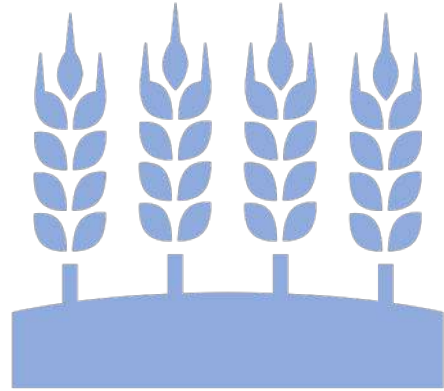


@etaagen

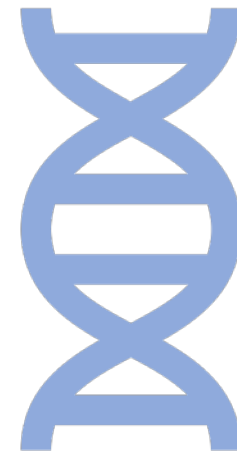
Roadmap to positional cloning



Fine-mapping



Reference genome



Sequencing



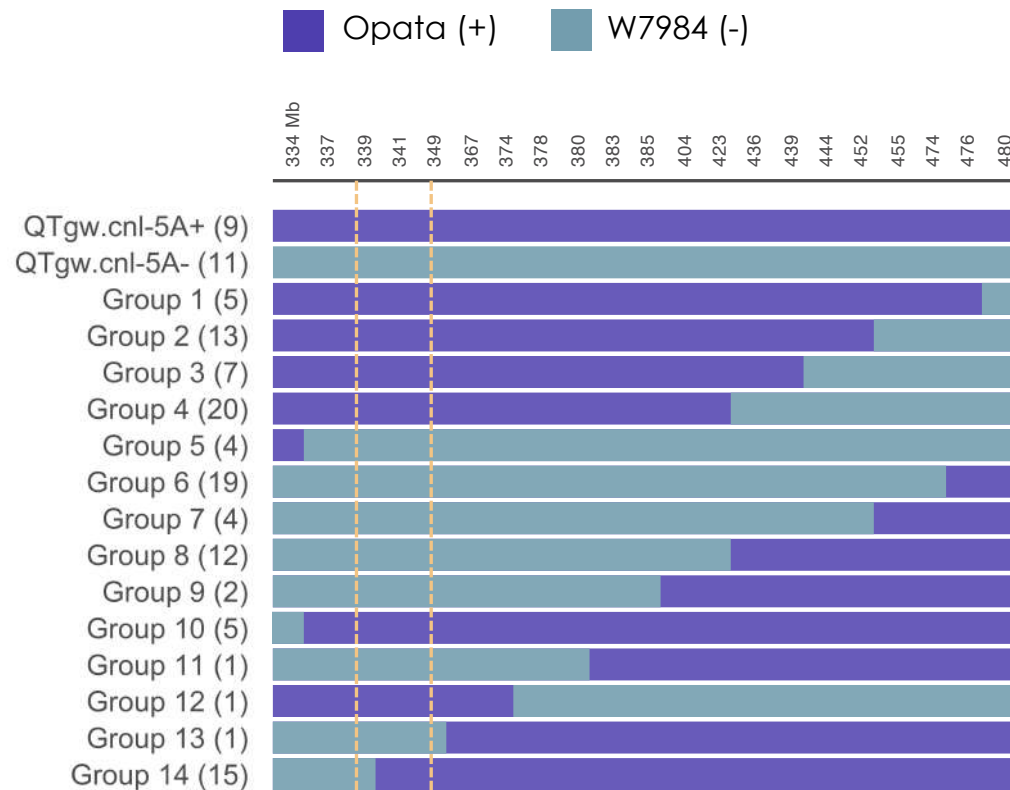
Functional validation

W7984 x Opata chromosome arm 5AL QTL associated with grain weight variation

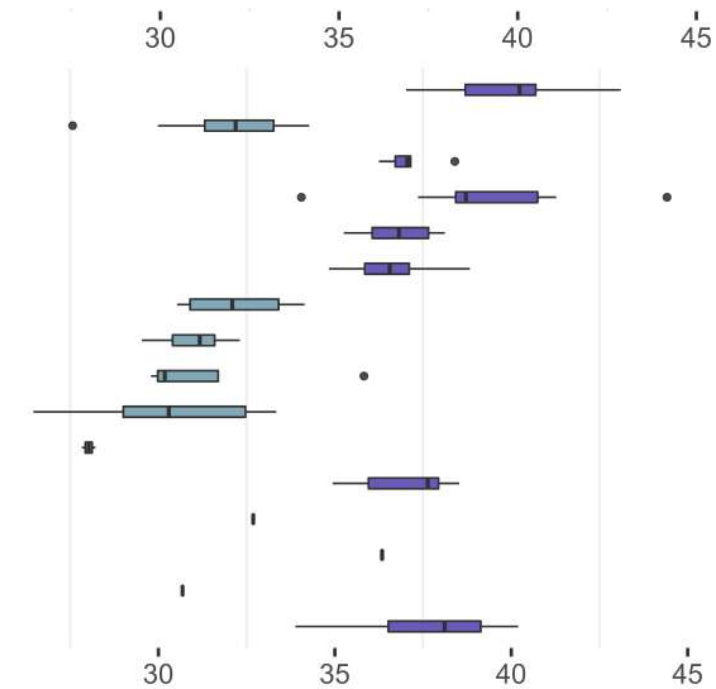
Genomic structural variations are **common** among polyploids

Fine-mapping population development

Heterogeneous inbred family (HIF): **109 F_{6:5}** lines with *QTgw.cnl-5A* recombination



Thousand grain weight (TGW, grams)

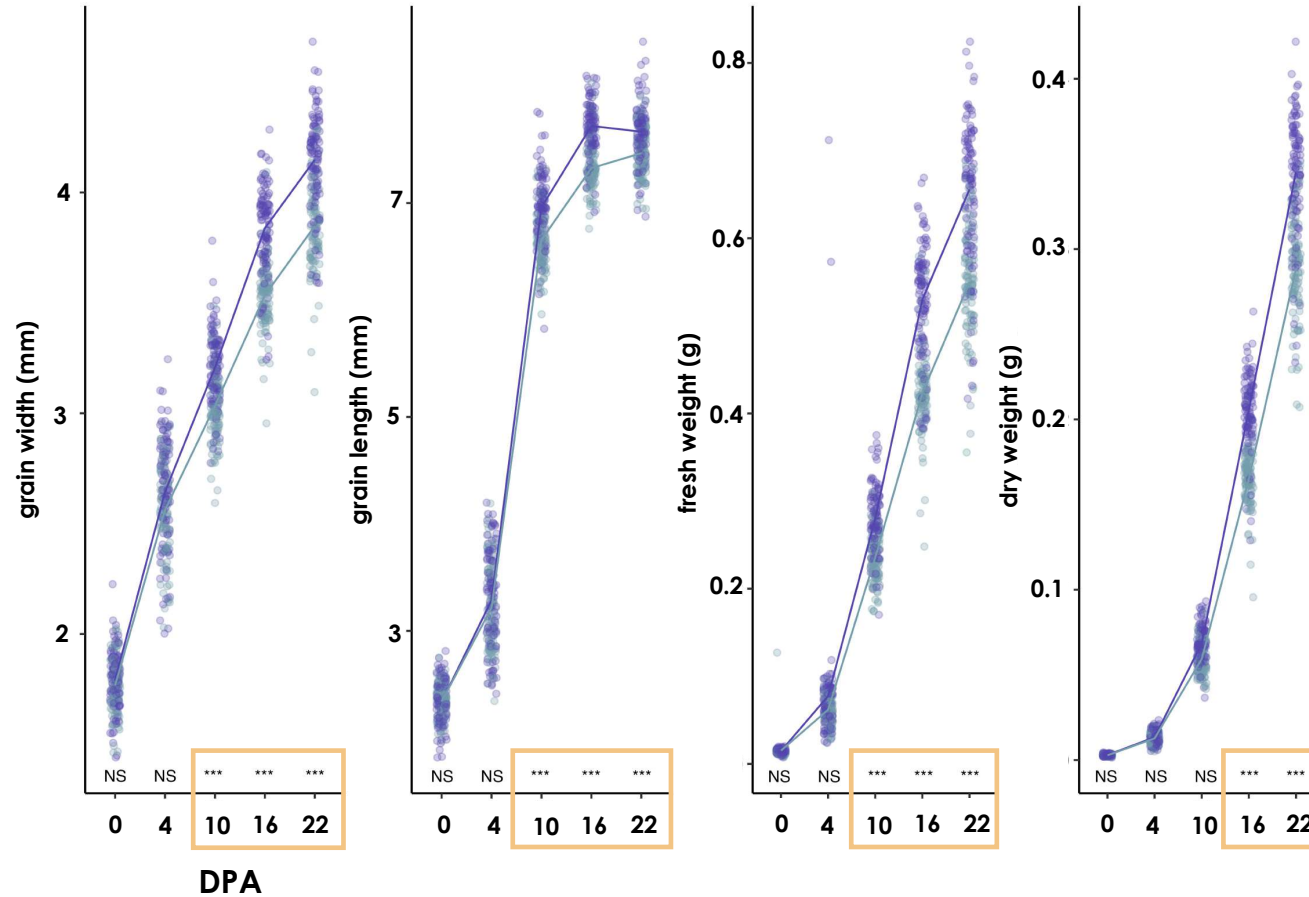


QTgw.cnl-5A+ **21.3%** heavier grains than *QTgw.cnl-5A-*

Variation in grain weight and morphology significantly associated with early grain development

QTgw.cnl-5A

- Opata (+)
- W7984 (-)



Bonferroni corrected p-value NS: non-significant, ***: $p \leq 0.001$

Gutierrez-Gonzalez et al., 2019:

SCIENTIFIC REPORTS

OPEN

Dense genotyping-by-sequencing linkage maps of two Synthetic W7984 × Opata reference populations provide insights into wheat structural diversity

Juan J. Gutierrez-Gonzalez¹, Martin Mascher^{2,3}, Jesse Poland⁴ & Gary J. Muehlbauer^{1,5}

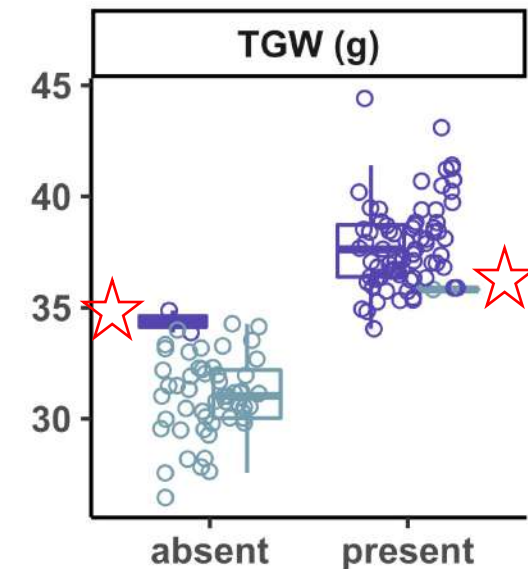
*“We found that chromosome arm 5AS is **missing** from W7984.”*

QTgw.cnl-5A is in linkage with chromosome arm 5AS structural variation

	<i>QTgw.cnl-5A</i> HIFs	<i>QTgw.cnl-5A+</i> Opata	<i>QTgw.cnl-5A-</i> W7984
Chr arm 5AS, absent	54	2	52
Chr arm 5AS, present	73	72	1

Correlation coefficient **0.91**

No significant interaction



***QTgw.cnl-5A* allele**



We used **RNA-seq** to investigate differentially expressed genes on chromosome arm 5AS and in the *QTgw.cnl-5A* candidate region

Chromosome arm 5AS present : *QTgw.cnl-5A+*

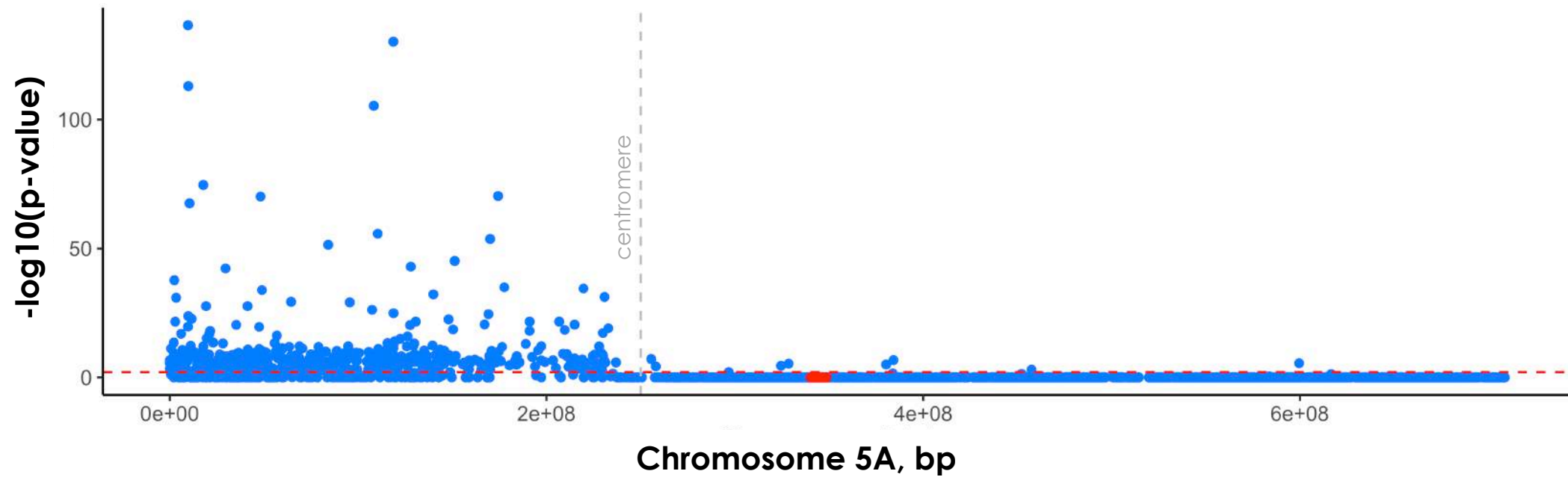
VS

Chromosome arm 5AS absent : *QTgw.cnl-5A-*

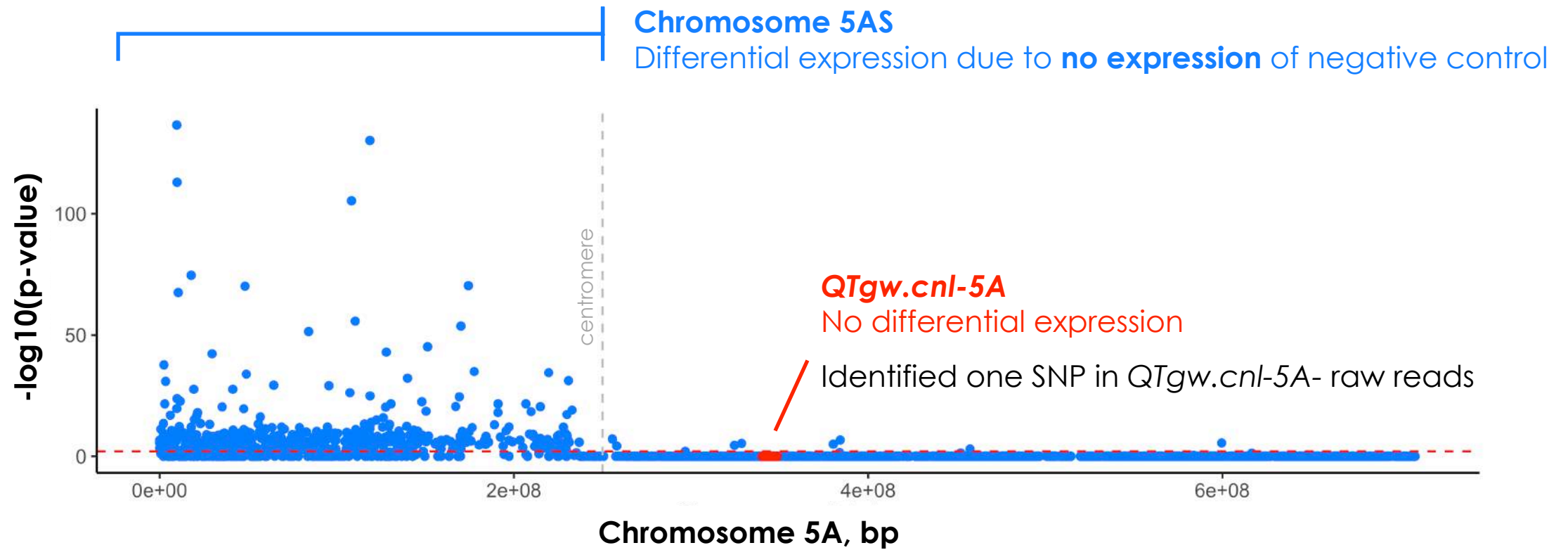
RNA-seq of HIFs confirmed the significance of chromosome arm 5AS structural variation

535 of the 556 differentially expressed genes are on chromosome 5A

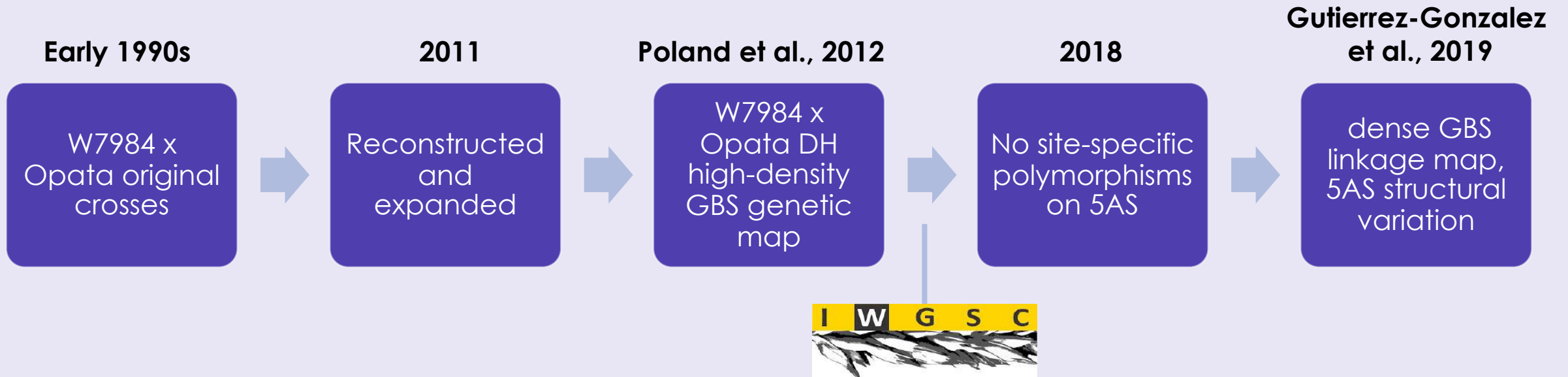
None of the homoeologous copies were differentially expressed



RNA-seq of HIFs confirmed the significance of chromosome arm 5AS structural variation



Detecting chromosome structural variation



Brinton et al., 2020 and Walkowiak et al., 2020, underscore the structural diversity of wheat

Polyploid positional cloning recommendations

1. Move from a SNP to haplotype-based approach to identify genetic diversity
2. Invest in sequencing to detect structural variants
3. Use the transcriptome to identify candidate genes
4. Traits with broad overlapping QTL may not be pleiotropic

Resources

Received: 25 January 2021 | Accepted: 19 April 2021

DOI: 10.1002/tpg2.20106

The Plant Genome 

ORIGINAL RESEARCH

Positional-based cloning ‘fail-safe’ approach is overpowered by wheat chromosome structural variation

Ella Taagen¹  | James Tanaka¹ | Alvina Gul² | Mark E. Sorrells¹

GitHub repository: github.com/etaagen/Taagen_2021_TPG

Acknowledgements

Dr. Mark Sorrells lab

Wheat CAP colleagues & collaborators

Eastern Regional Small Grains Genotyping Lab

International Wheat Genome Sequencing Consortium

Open-source tools



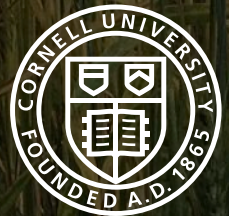
Challenges and opportunities in positional cloning and structural variation in polyploid crops

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Sources

Taagen, E., Tanaka, J., Gul, A., & Sorrells, M. E. (2021). Positional-based cloning 'fail-safe' approach is overpowered by wheat chromosome structural variation. *Plant Genome*. 2021;14:e20106. <https://doi.org/10.1002/tpg2.20106>

Gutierrez-Gonzalez, J.J., Mascher, M., Poland, J. *et al.* Dense genotyping-by-sequencing linkage maps of two Synthetic W7984×Opata reference populations provide insights into wheat structural diversity. *Sci Rep* **9**, 1793 (2019). <https://doi.org/10.1038/s41598-018-38111-3>

Brinton, J., Ramirez-Gonzalez, R. H., Simmonds, J., Wingen, L., Orford, S., Griffiths, S., Haberer, G., Spannagl, M., Walkowiak, S., Pozniak, C., & Uauy, C. (2020). A haplotype-led approach to increase the precision of wheat breeding. *Communications Biology*, **3**, 712. <https://doi.org/10.1038/s42003-020-01413-2>

Walkowiak, S., Gao, L., Monat, C., Haberer, G., Kassa, M. T., Brinton, J., Ramirez-Gonzalez, R. H., Kolodziej, M. C., Delorean, E., Thambugala, D., Klymiuk, V., Byrns, B., Gundlach, H., Bandi, V., Siri, J.N., Nilsen, K., Aquino, C., Himmelbach, A., Copetti, D., ... Pozniak, C. J. (2020). Multiple wheat genomes reveal global variation in modern breeding. *Nature*, **588**, 277–283. <https://doi.org/10.1038/s41586-020-2961-x>