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## Presence/absence variations and SNPs equally contribute to the variations of protein and metabolite abundance

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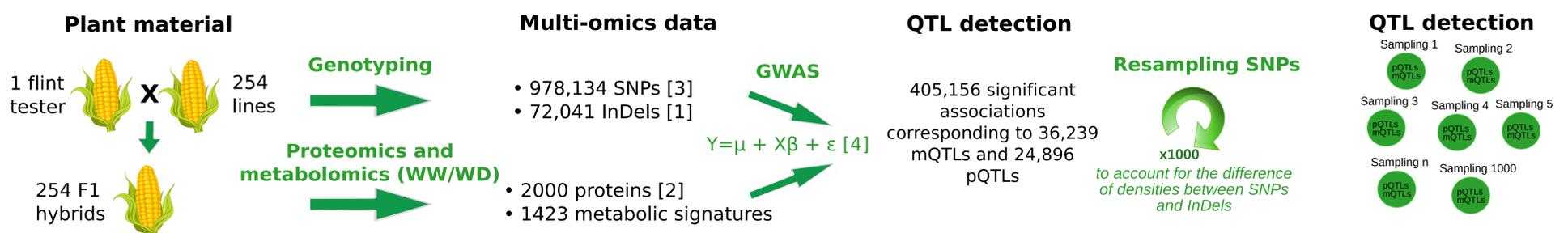
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## Introduction

Presence Absence Variations (PAVs) are structural polymorphisms of the genome that can be induced by insertions or deletions of DNA sequences (InDels). Because InDels can occur in coding sequences or in gene regulatory regions, they are supposed to be involved in adaptation to the environment. However, their contribution to the genetic determinism of traits and genotype by environment (GxE) interactions remains largely unknown. Recently, Mabire et al. [1] published InDel genotyping data for a diversity panel of Dent maize lines. In addition, leaves samples taken on hybrids obtained from this same panel and grown under two watering conditions (well-watered, WW, and water deficit, WD) were analysed by proteomics [2] and metabolomics (unpublished). In this study, we took advantage of these data to evaluate to which extent InDels contribute to the abundance variations of proteins and metabolites as compared to SNPs. To address this issue, we performed genome-wide association studies (GWAS) to detect quantitative trait loci for protein abundance (pQTLs) and metabolite abundance (mQTLs) from both InDels and SNPs.

## Material and methods



## InDels allow to detect new QTLs

Compared to SNPs, InDels allowed to detect 2061 and 2705 new pQTLs and mQTLs, respectively (Fig. 1).

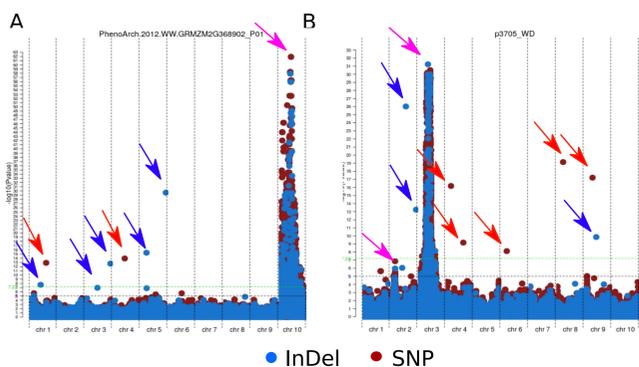


Fig. 1 : Manhattan plot obtained for (A) the protein "GRMZM2G358902" and (B) the metabolite signature "p3705". QTLs can be determined by InDels only (blue arrows), SNP only (red arrows) or both (pink arrows).

## InDels and SNPs have similar effects

The proportion of variance explained by InDels and SNPs is similar in the two watering condition and for both proteins and metabolites (Fig. 2).

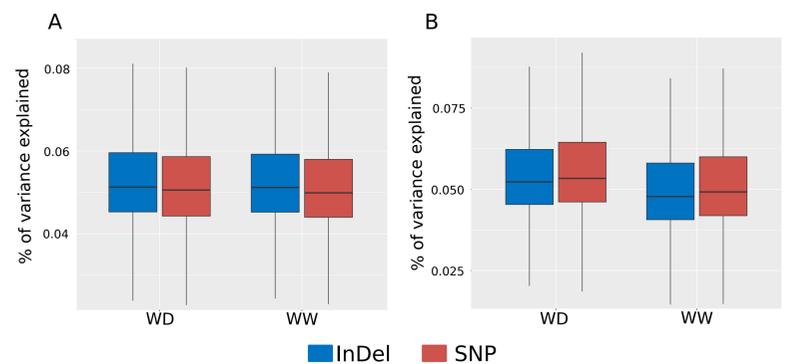


Fig. 2 : Proportion of variance explained by (A) pQTLs and (B) mQTLs in the two watering conditions

## SNP and InDels have the same power of QTLsdetection

SNPs and InDels allowed to detect equivalent proportions of pQTLs and mQTLs (Fig. 3). In addition, water deficit had no influence on the number of pQTLs and mQTLs detected (Table 1).

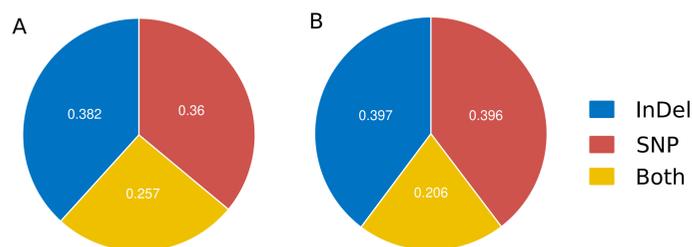


Fig. 3 : Proportion of (A) pQTLs and (B) mQTLs detected by InDels, SNPs or both. Result obtained after re-sampling the SNPs.

		WW	WD
# of pQTLs detected by	InDels	3090	3101
	SNPs	3050	3196
# of mQTLs detected by	InDels	2046	2324
	SNPs	2062	2254

Table 1 : Number of pQTLs and mQTLs detected by InDels and SNPs according to the watering condition. The QTLs detected by the two types of polymorphism were assigned to the one that was the most strongly associated to the molecular phenotype.

## Conclusions and perspectives

This study shows that InDels can be associated with the variations of molecular traits and some of these associations are not detected by SNPs. It is therefore very relevant to take them into account in further genetics studies. InDels and SNPs equally contributed to the variation of molecular traits. However, since molecular phenotyping was performed on hybrids, the effect of InDels may be hidden by complementation with the tester. Further work will focus on the analysis of the functions of the proteins associated to pQTLs that determined by InDels.

[1] Mabire et al. 2019, BMC Genomics 20, no. 1 (December 2019): 848. <https://doi.org/10.1186/s12864-019-6136-9>  
 [2] Blein-Nicolas et al. 2020, Genome Research 30, no. 11 (November 2020): 1593–1604. <https://doi.org/10.1101/gr.255224.119>  
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 [4] Yu et al. 2006, Nat Genet 38 (2006): 203–8,