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GWAS to post-GWAS: in the context of lung cancer study

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Background and Aim

- Lung cancer (LC) is the second most common cancer and the leading cause of cancer death worldwide, accounting for 18% of the total cancer deaths.
- Genome-wide association studies (GWAS) have identified approximately 45 genomic loci with a large number of genetic variants that are significantly associated with LC risk.
- many novel computational and experimental tools now became available to accelerate the functional assessment of lung cancer-associated variants
- but the biological mechanisms underlying these associations remain largely unknown due to th e lack of enough functional study so far
- The aim of this study to explore the current status of molecular insights from GWAS to post-GW AS era.

Methods & Results

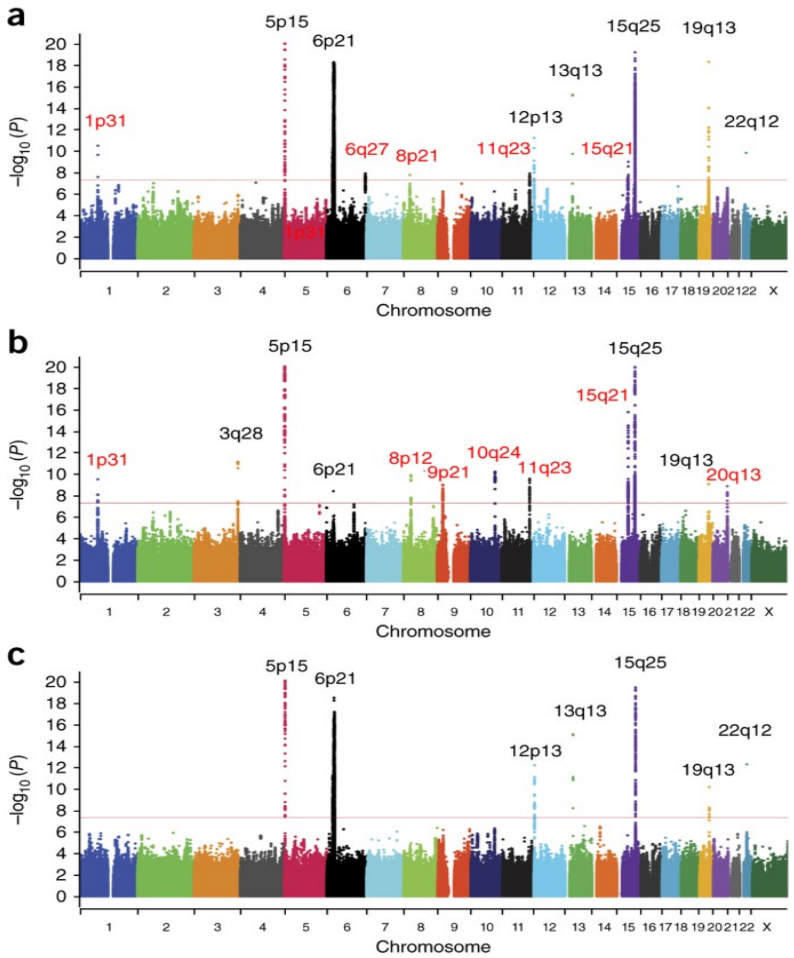

Figure 1: Manhattan plots of lung cancer risk overall and by histological subtype

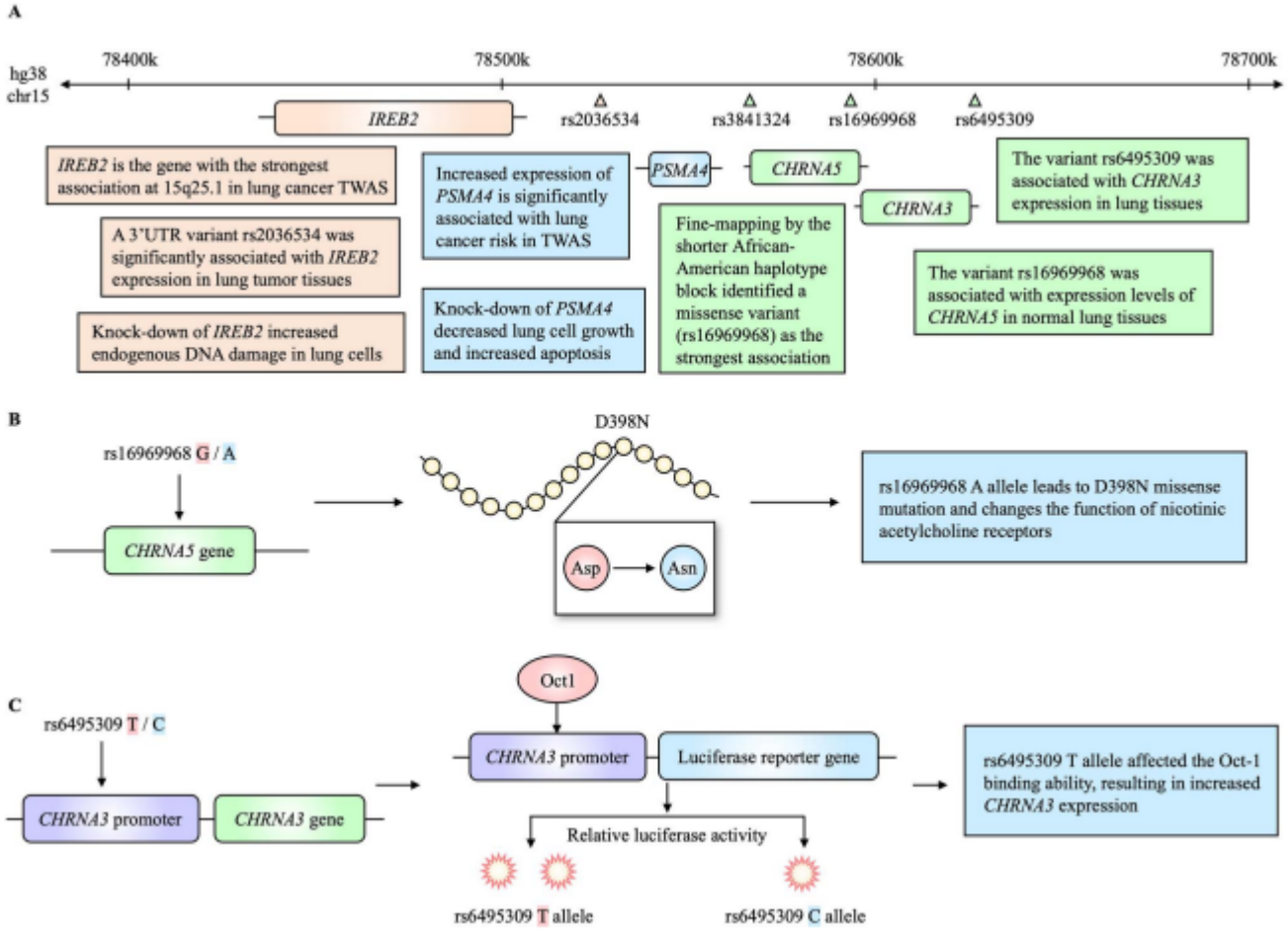

Figure 2. Summary of the functional findings from lung cancer GWAS locus at 15q25.1.

Cont….

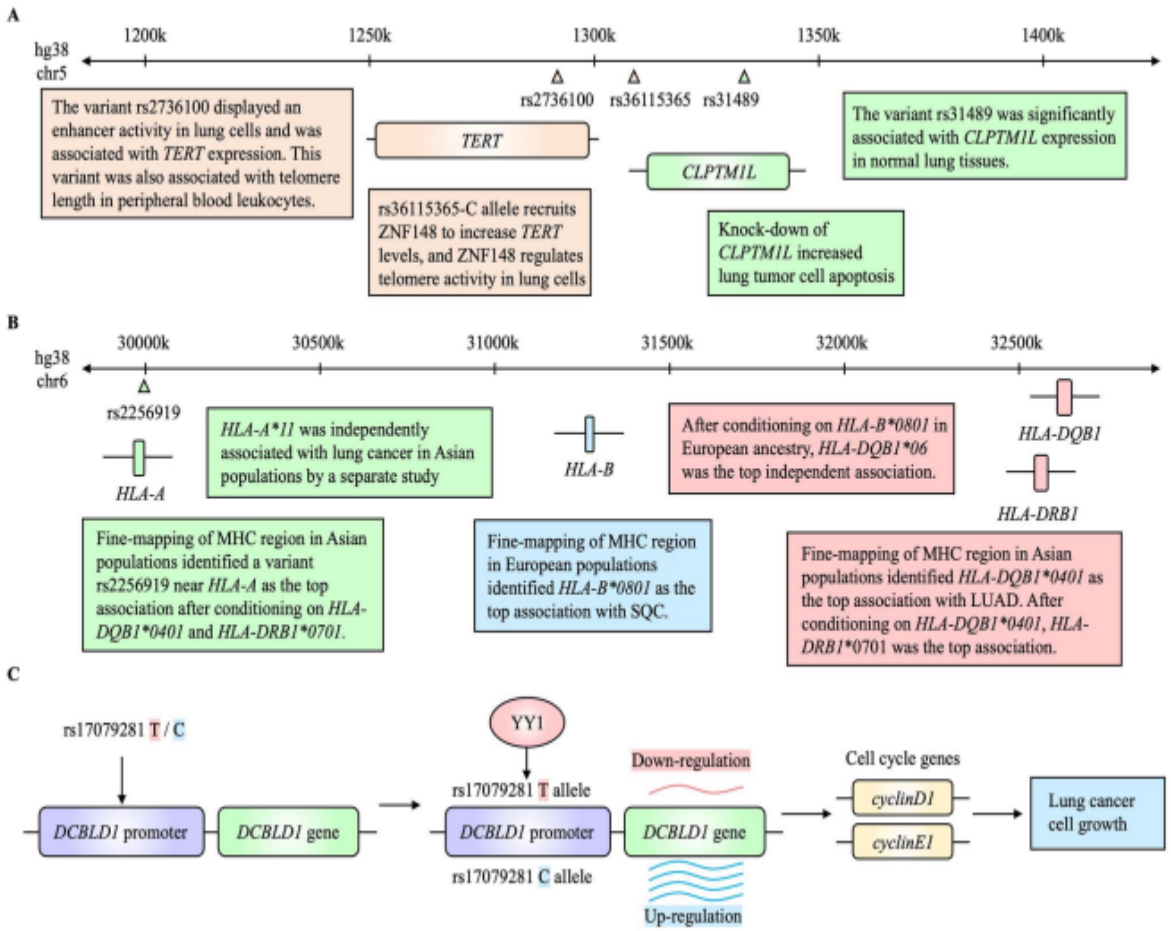

Figure 3. Summary of the functional findings from lung cancer GWAS loci at 5p1 5.33, 6p21 (MHC) and 6p22.1.

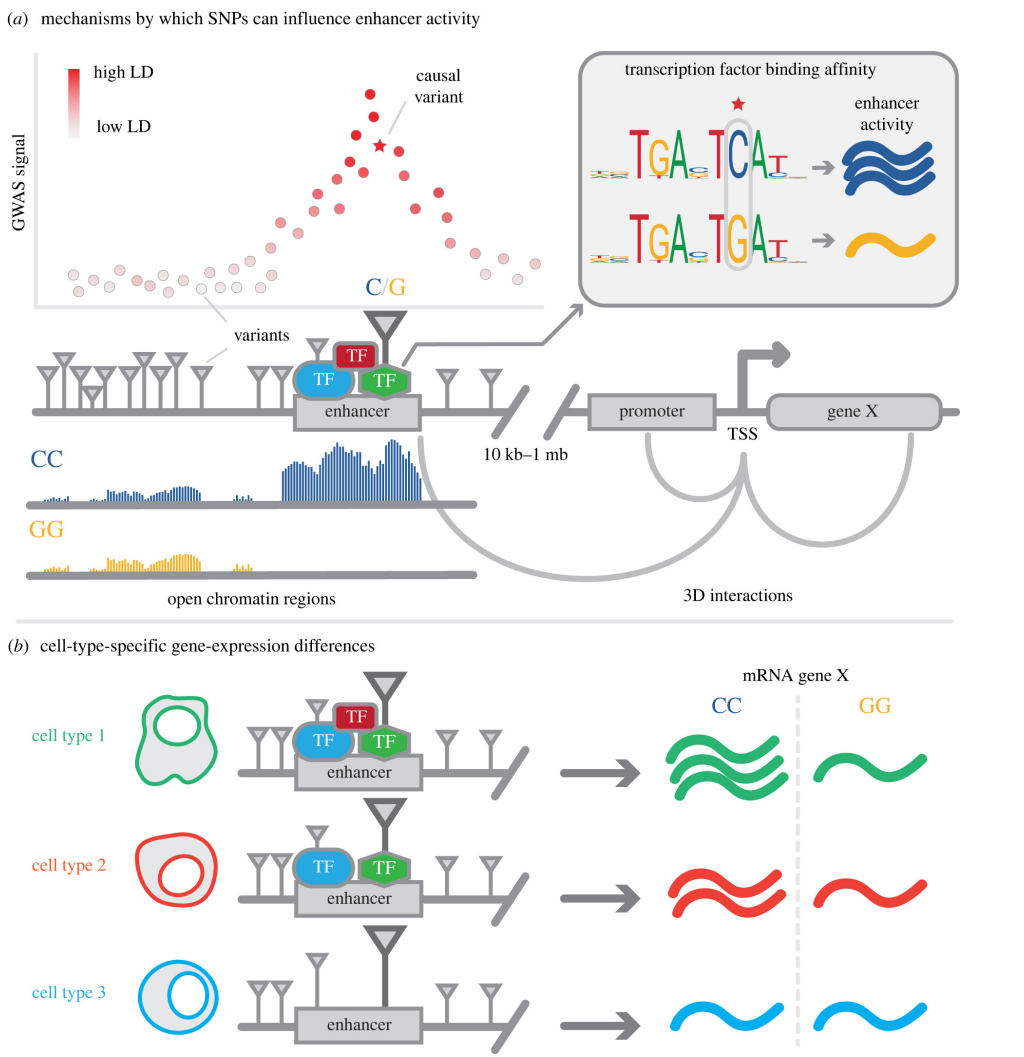

Figure 4. A practical view of fine-mapping and gene prioritization in the post-ge nome-wide association era

Cont….

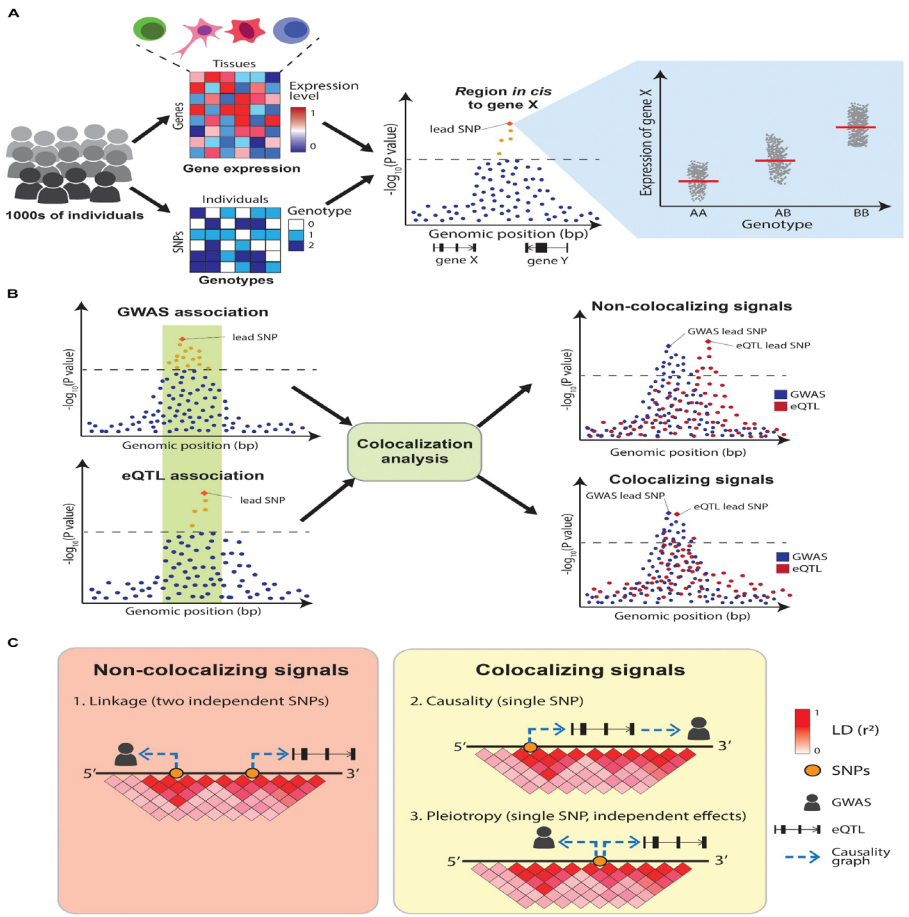

FIGURE 5. Overview of eQTL-mapping and colocalization.

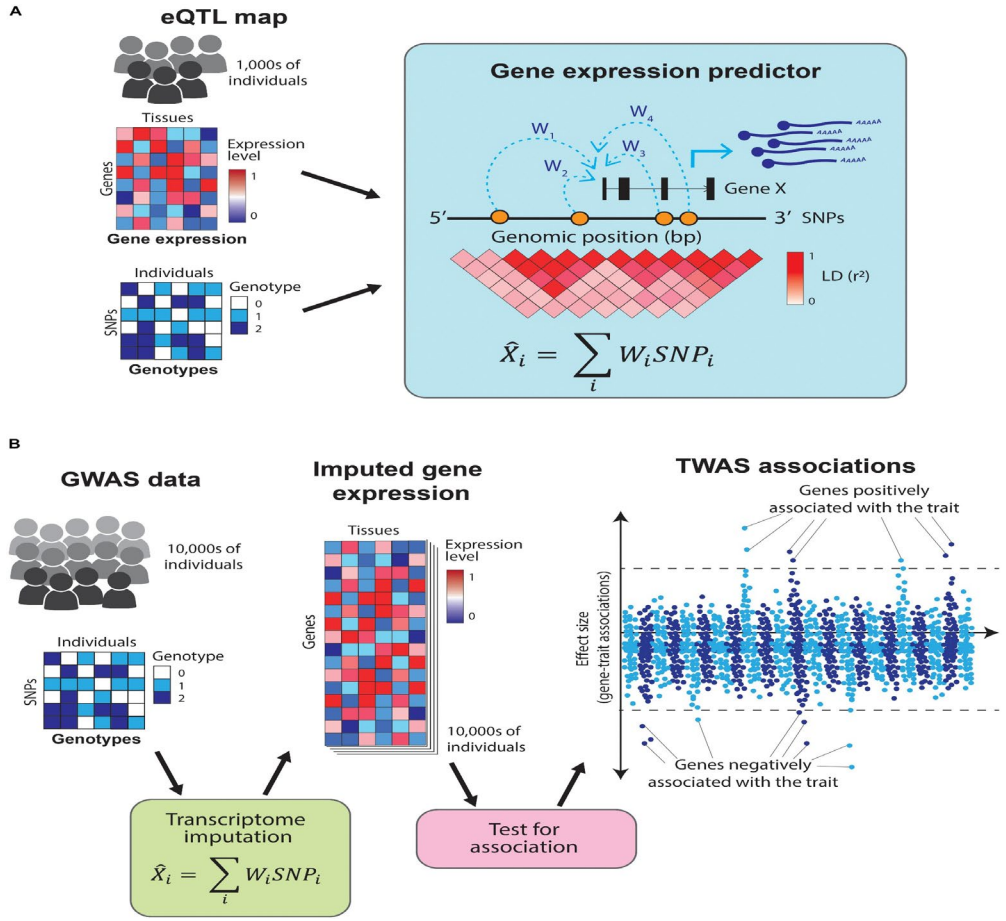

FIGURE 6. Overview of transcriptome-wide association studies TWAS leverage information from eQTL catalogs and GWAS studies to directly associate traits to genes.

Conclusion

- Still these techniques are not enough to know the molecular mechanism of genetic variants over the target genes.
- Therefore, it is necessary to study the function of causal genetic variants of LC on their target genes to get the highest benefits in the long run.

Thank you

All