

Characterization and predictive role of Human-specific genes in Acute Lymphoblastic Leukemia

Gloria Lugoboni [1]*, Lorenzo Santarelli [1]*, Thomas Sirchi [1]*, Andrea Tonina [1]*, Matteo Ganesello [1], Federica Ressa [1], Emma Busarello [1], Valter Cavecchia [3], Luca Tiberi [1], Toma Tebaldi [1], Enrico Blanzieri [2]

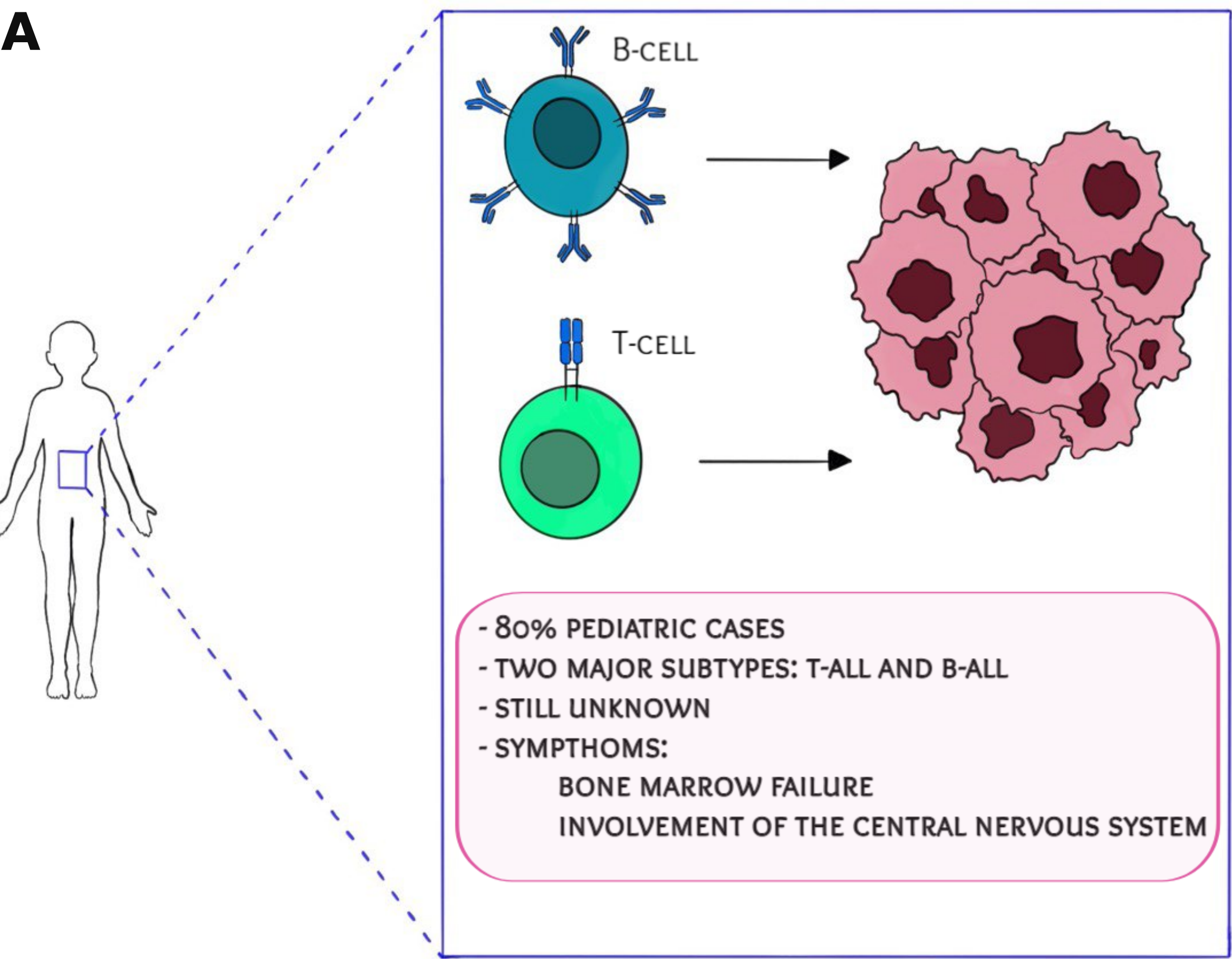
[1] Department of Cellular, Computational and Integrative Biology (CIBIO), University of Trento, via Sommarive n. 9, 38123 Povo, Trento, Italy.

[2] Department of Information Engineering and Computer Science, University of Trento, Via Sommarive 9, 38123 Povo, Trento, Italy

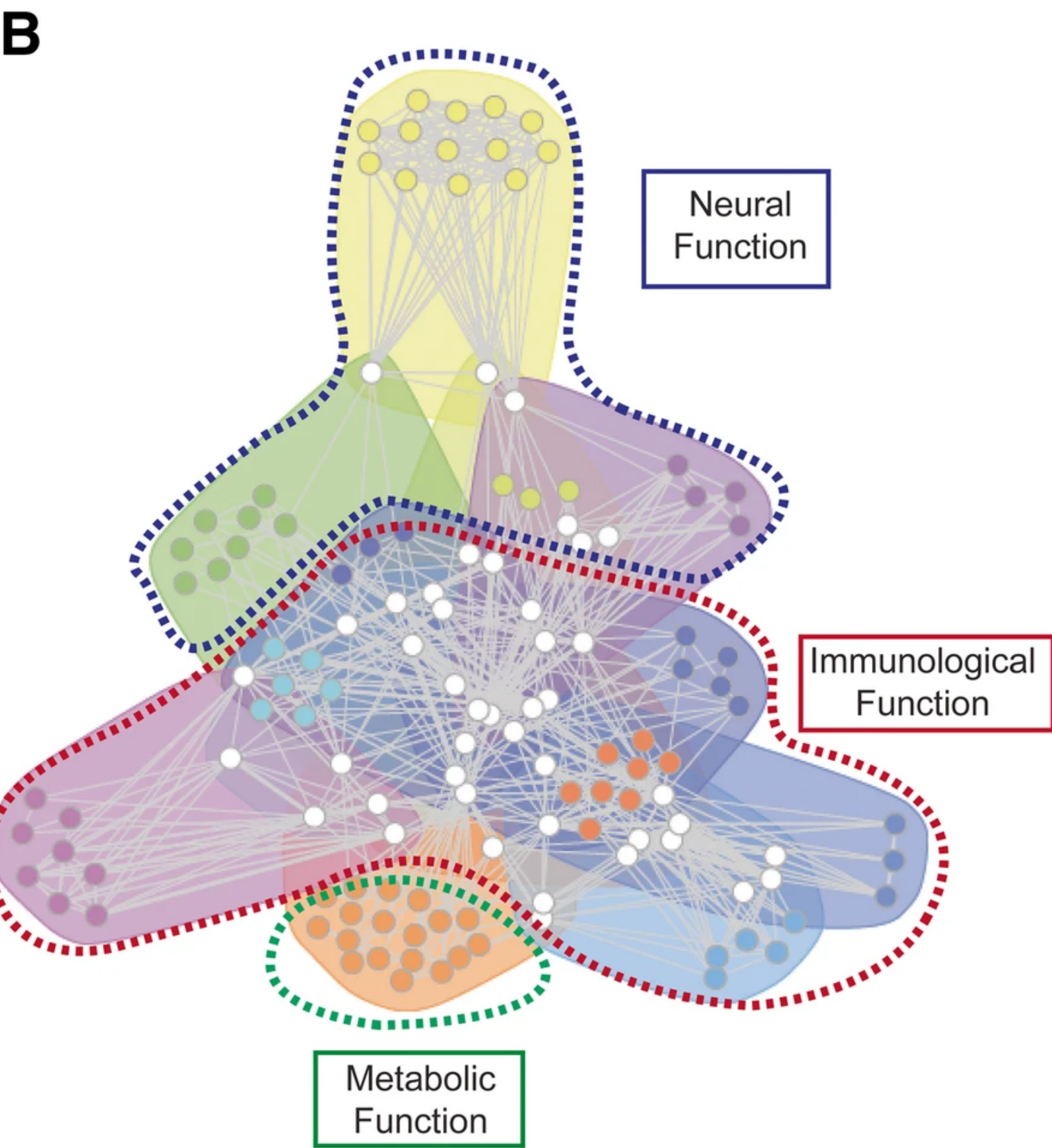
[3] Institute of Materials for Electronics and Magnetism (CNR), via alla Cascata 56/c, 38123, Povo, TN, Italy

* These authors contributed equally

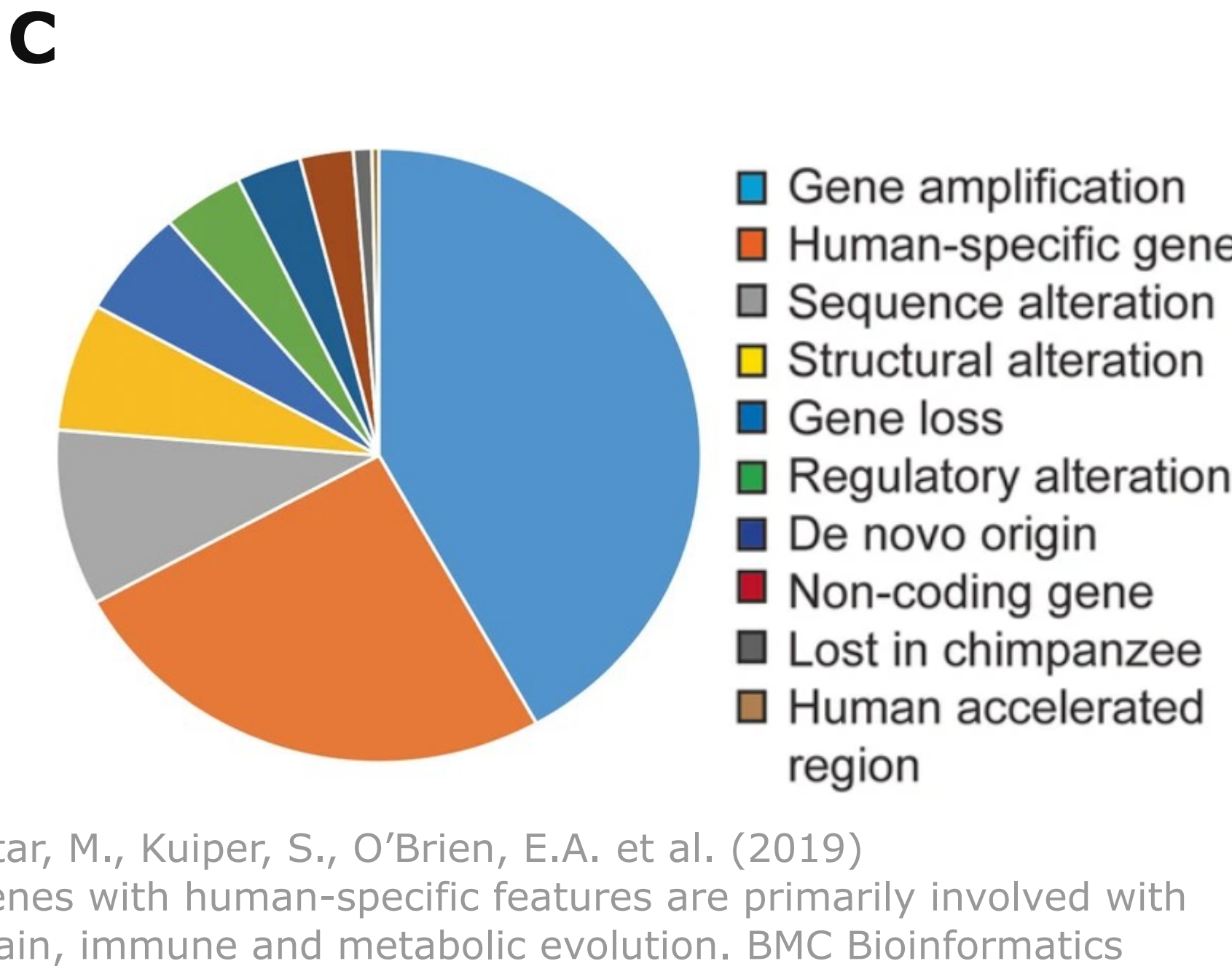
Overview of Acute Lymphoblastic Leukemia (ALL)



Main functions associated with human-specific genes

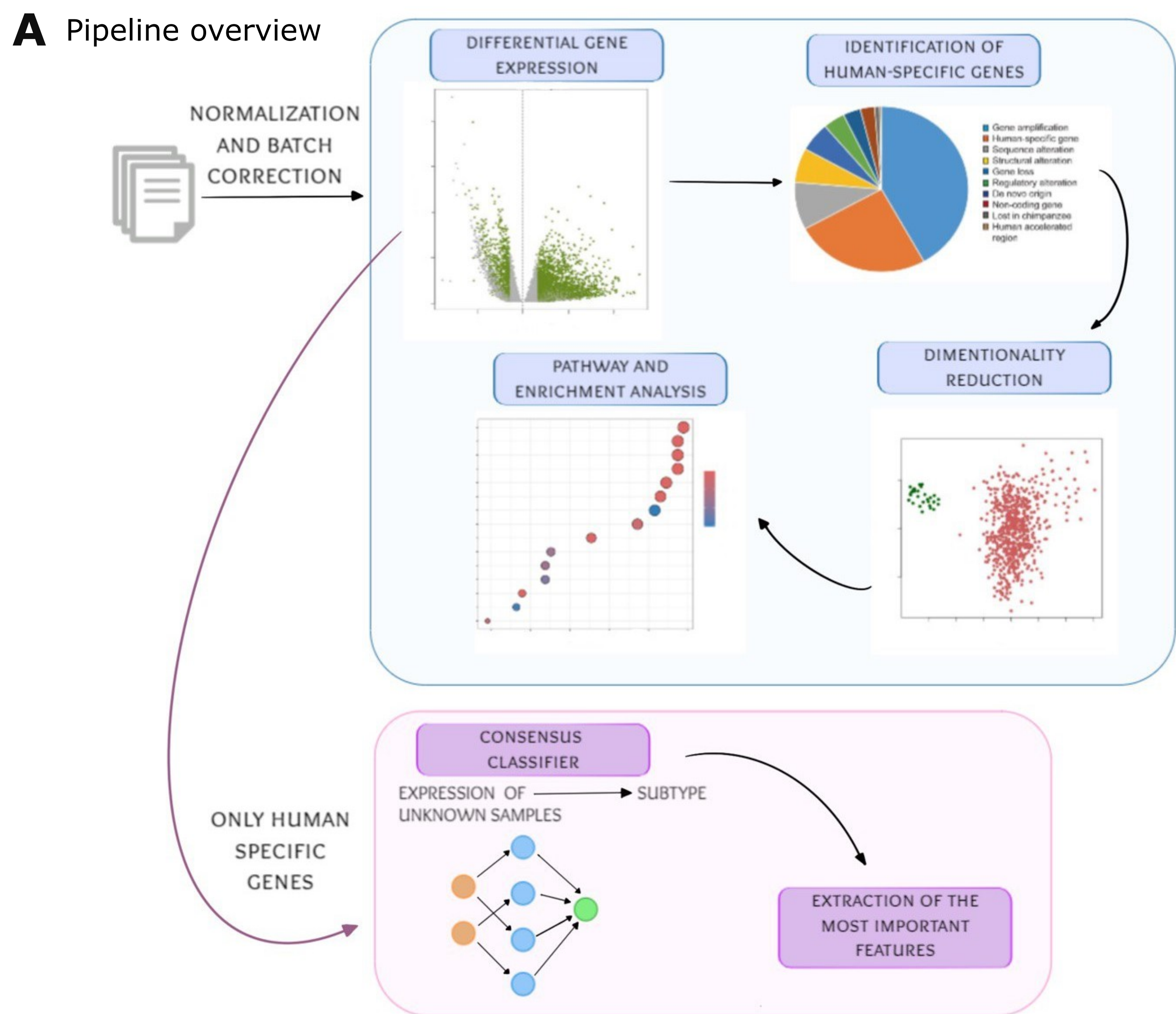


Mechanism of origin of known human-specific genes



We talk of "human-specific" features, or human-specific (HS) genes, to indicate the main differences that can be found at the genetic level between humans and our close relatives, the chimpanzees.

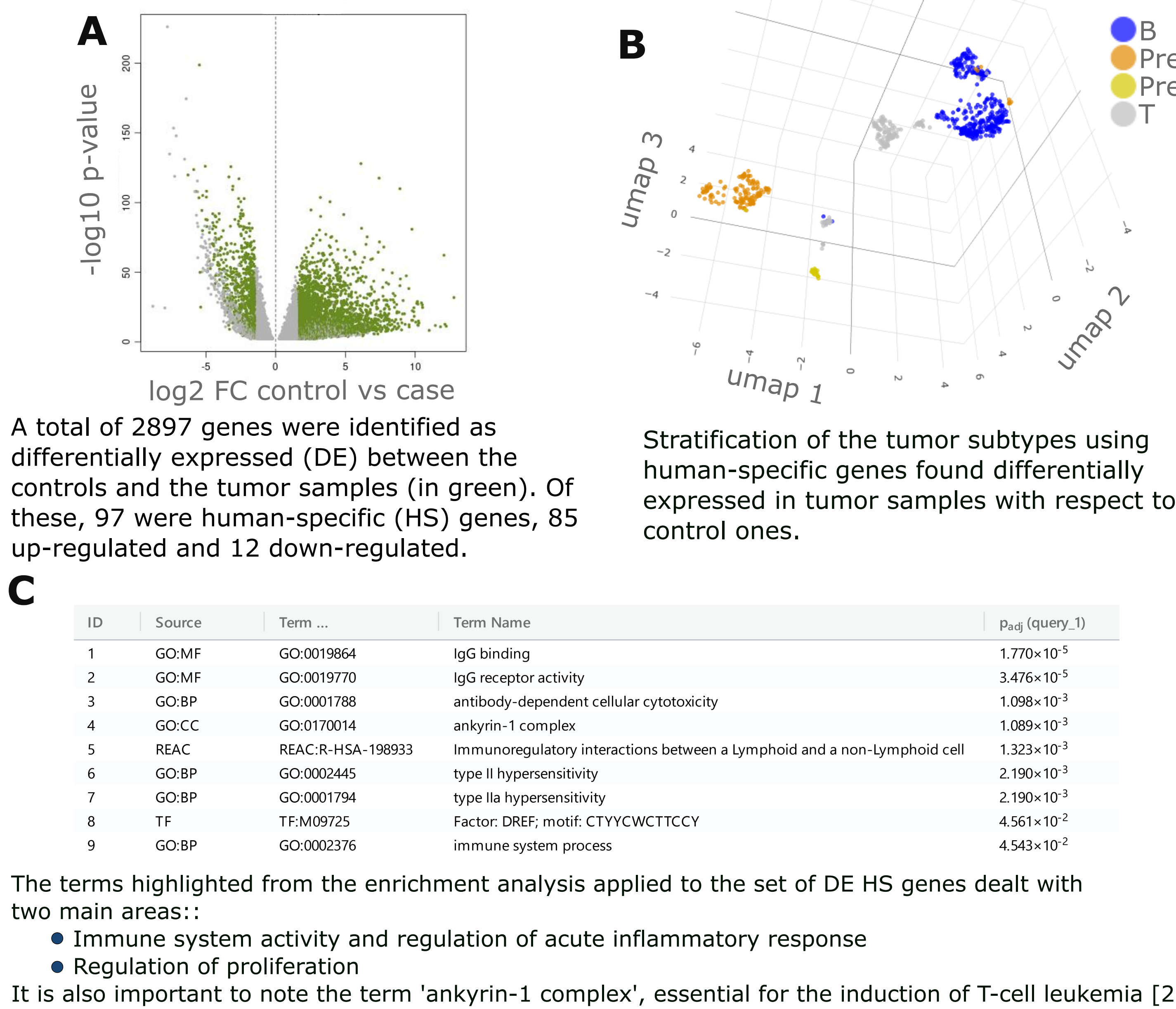
Comprehensive Analysis of ALL via HS genes Profiling



B Datasets overview

Dataset	Origin	Control samples	Tumor samples	Age
GSE84445	Blood	20	0	Unknown
GSE133499	Both	0	42	Pediatric
GSE181157	Both	0	173	Pediatric
GSE227832	Both	10	330	Pediatric
Cohort_7_8	Unknown	0	107	Both
GSE139073	Bone marrow	40	0	Adult
GSE162562	Blood	5	0	Unknown
GSE115736	Blood	18	0	Unknown

Unraveling ALL using Human-specific genes



Conclusions

- Among the retrieved differentially expressed genes, it was possible to identify human-specific genes stratifying the tumor and the various subtypes (B, T, PreB and PreT).
- Enrichment analysis suggests that the human-specific genes are involved in immune system activity, its regulation and proliferation, all relevant processes associated with cancer.
- Through the differentially expressed human-specific genes we were able to create a consensus classifier capable of associating unknown data to specific tumor subtypes.
- Among human-specific genes with higher importance for our models, we found EBF1, a cancer-related gene connected to the signal transduction in leukemia [3], MYO7B, a known proto-oncogenic driver [4] and RAB6C, a member of the RAS oncogene family [5].

References

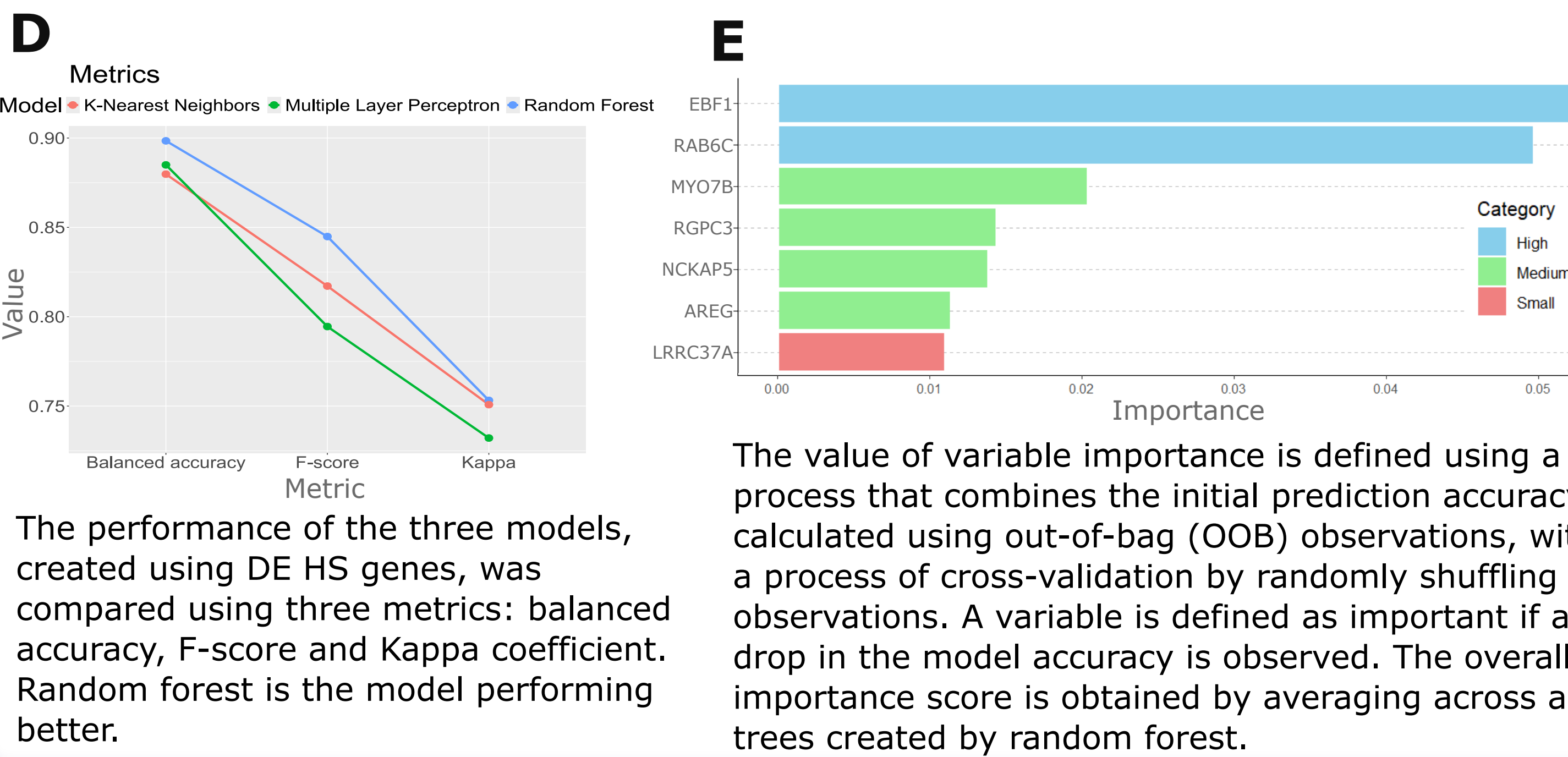
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Future perspectives/Outlooks

- Ampliate our data and effectuate the analysis on a bigger dataset to gain more statistical significance.
- Further analyses of the differences (and similarities) between the different ALL subtypes should be performed. The same can be said about the characterization of pediatric and adult samples.
- Further analyses regarding the genetics of human-specific genes are needed, common SNPs/SNVs in these genes should be analyzed to further understand the impact of such variants in the context of ALL.



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