

# Genomic and transcriptomic analyses of desmoid tumor reveals enrichment of transforming growth factor beta responsive signature

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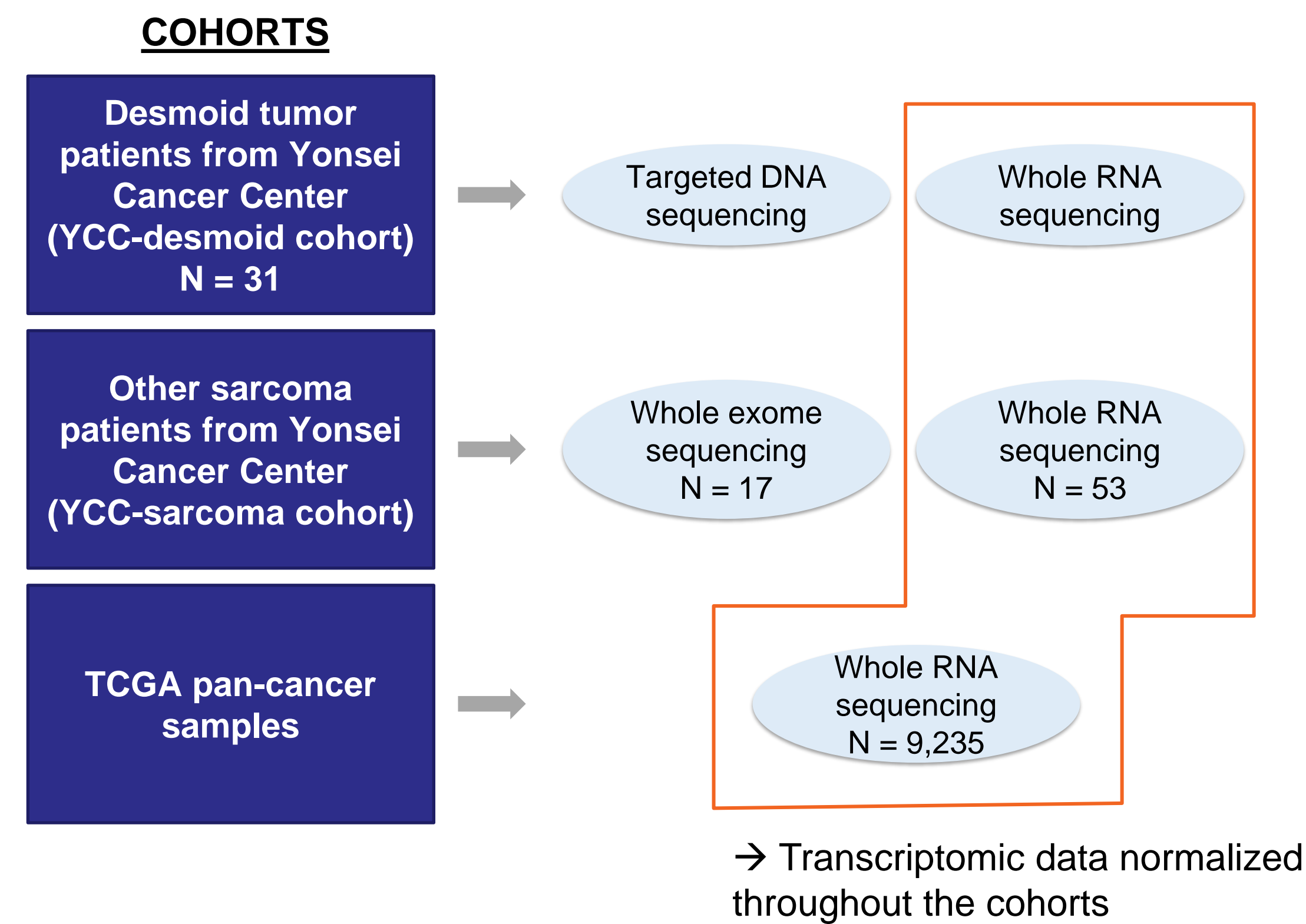
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## BACKGROUND

- **Desmoid tumor** (desmoid-type fibromatosis) is a rare aggressive tumor where there is no satisfying systemic treatment
- Transforming growth factor-β (TGF-β) expression is increased in the desmoid tumor
- Hence, inhibition of TGF-β response may be a potentially effective therapeutic strategy in desmoid tumor
- **We analyzed genomic and transcriptomic data of tumor samples from patients with desmoid tumor**

## MATERIALS and METHODS

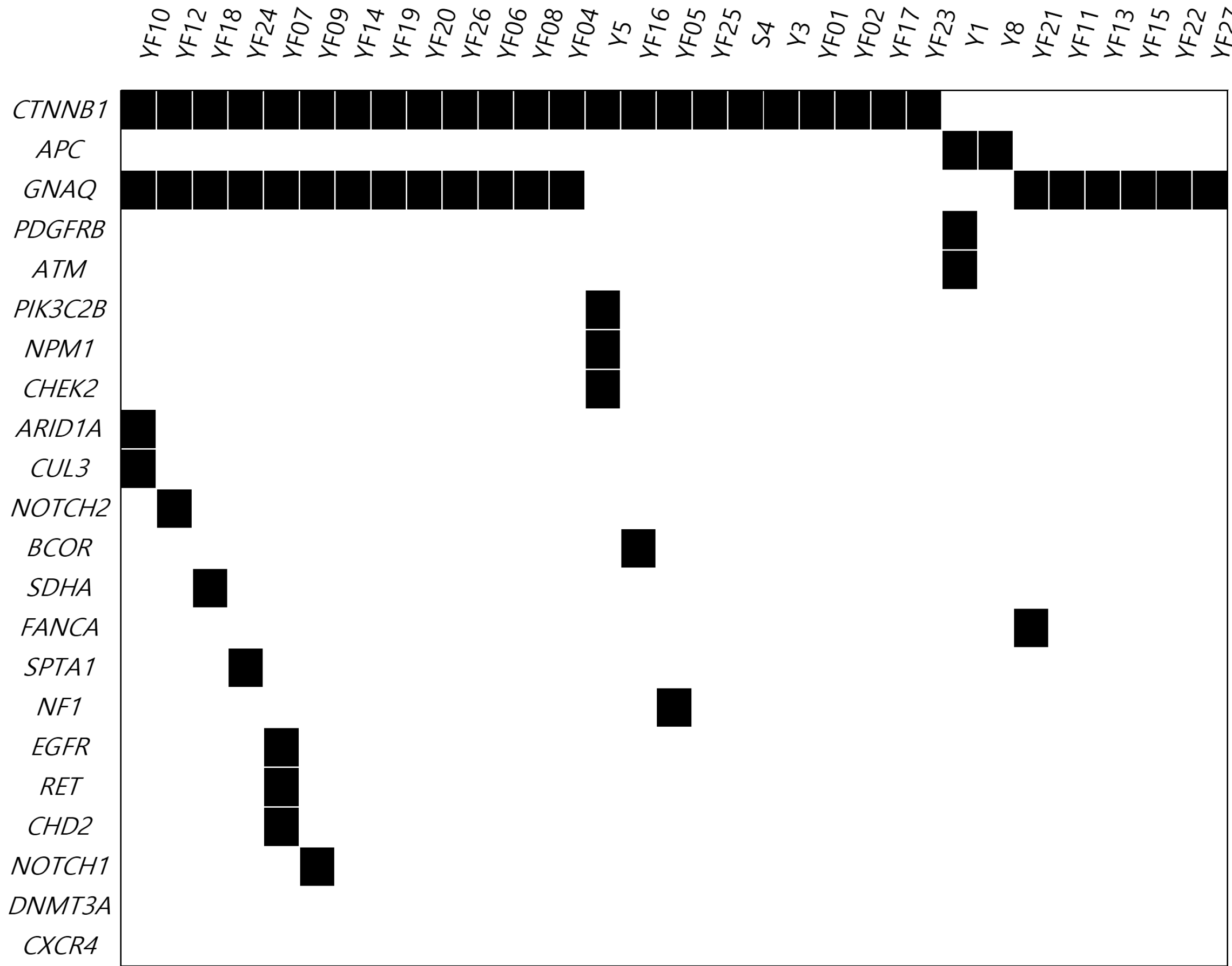
Figure 1. Schematic flow of materials and methods



- To evaluate the enrichment of TGF-β responsive signature (TBRS) in tumor and microenvironment, we calculated the mean expression values of fibroblast-TBRS (F-TBRS) from previous study.<sup>1</sup>

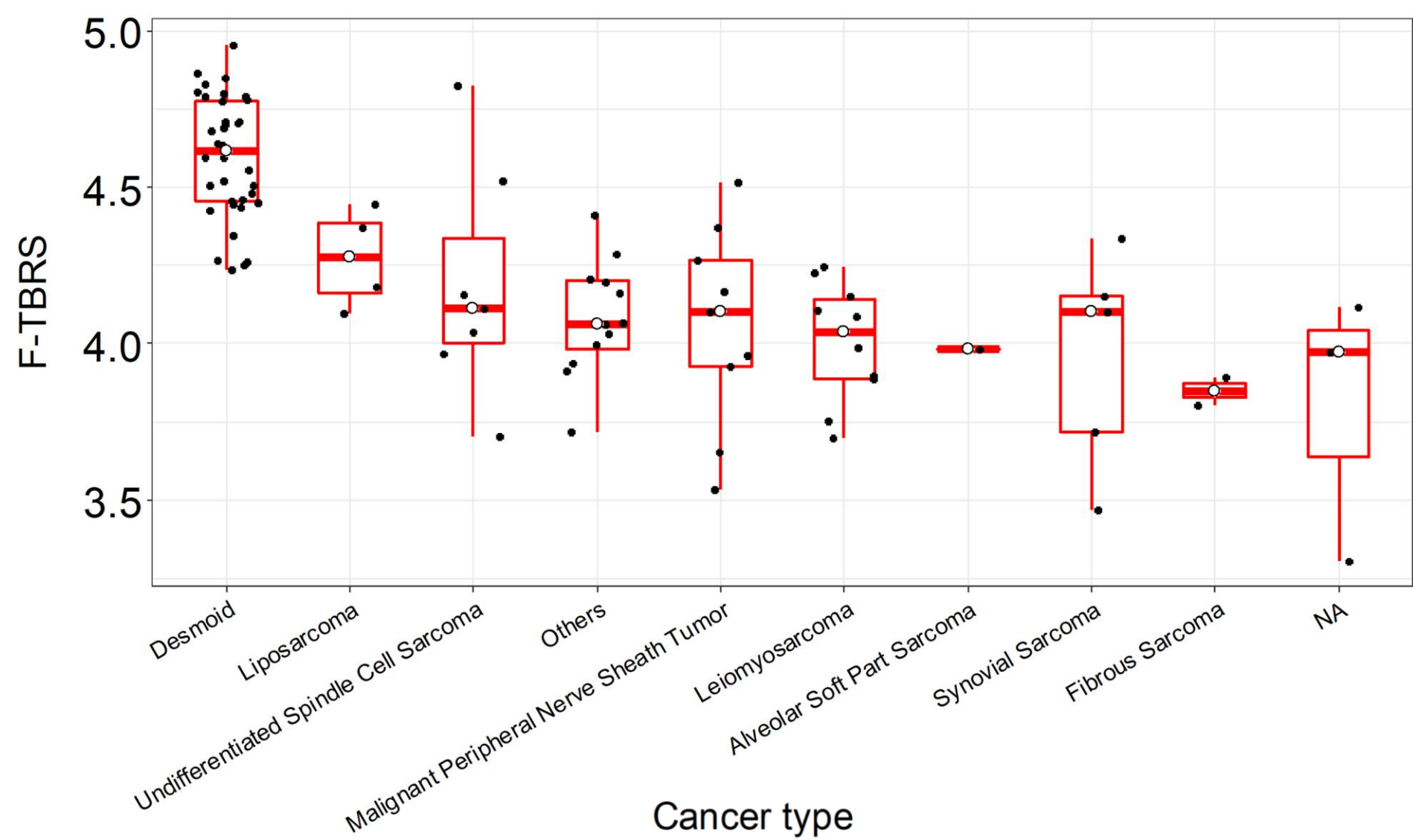
## RESULTS

Figure 2. Genomic landscape of desmoid tumor



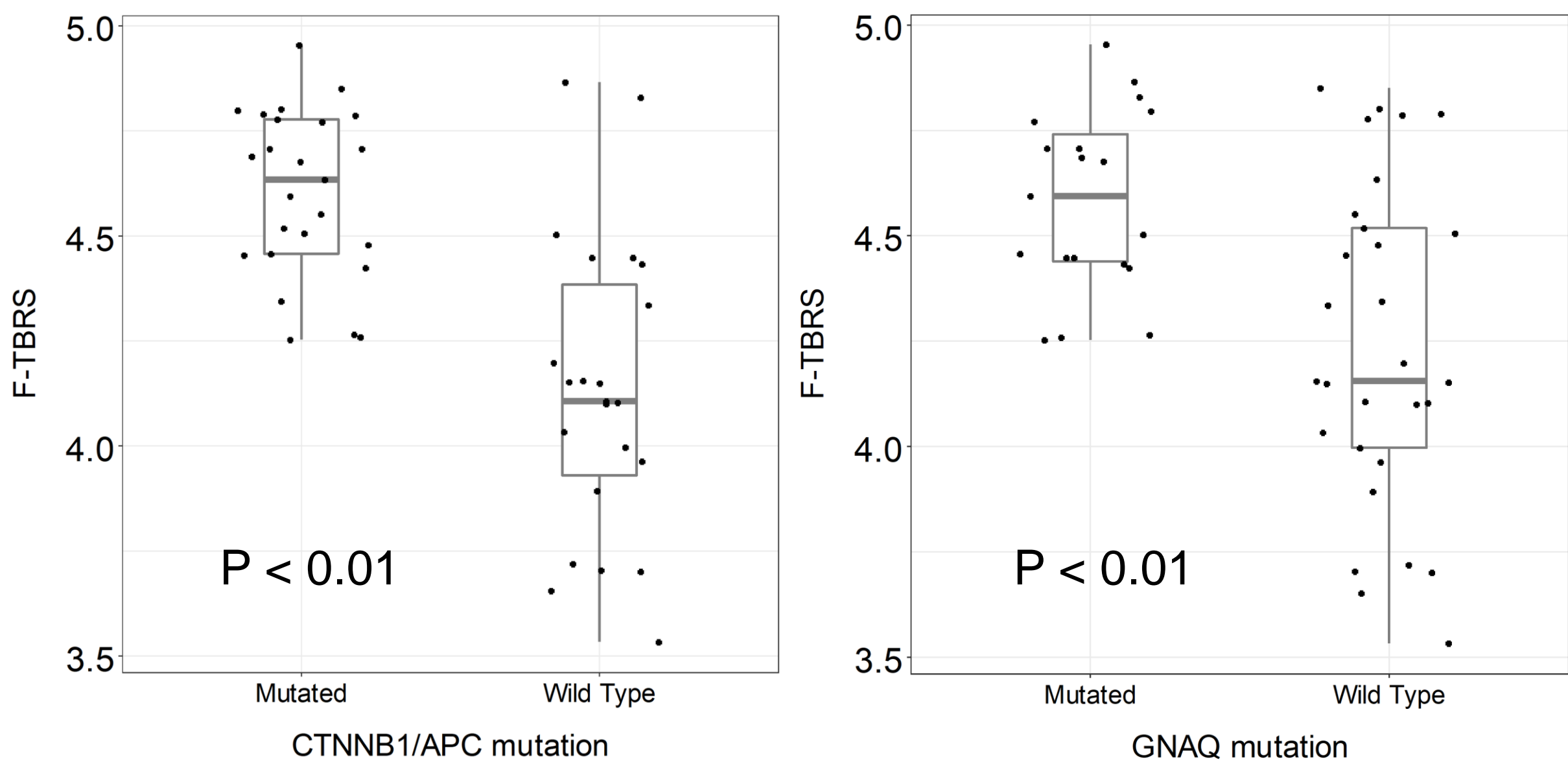
- *CTNNB1*, *GNAQ*, and *APC* mutations were the top 3 frequent mutations.
- All desmoid tumor tissue samples harbored **at least one** of *CTNNB1*, *APC*, *GNAQ* mutation

Figure 3. F-TBRS comparison among the various cancer types



- The boxplots above show the F-TBRS levels of YCC-desmoid and YCC-sarcoma cohorts.
- Desmoid tumor samples showed the highest F-TBRS.

Figure 4. F-TBRS comparison according to mutation status in YCC-desmoid and YCC-sarcoma patients



- *CTNNB1/APC* mutation and *GNAQ* mutation were both associated with higher enrichment in F-TBRS

## CONCLUSIONS

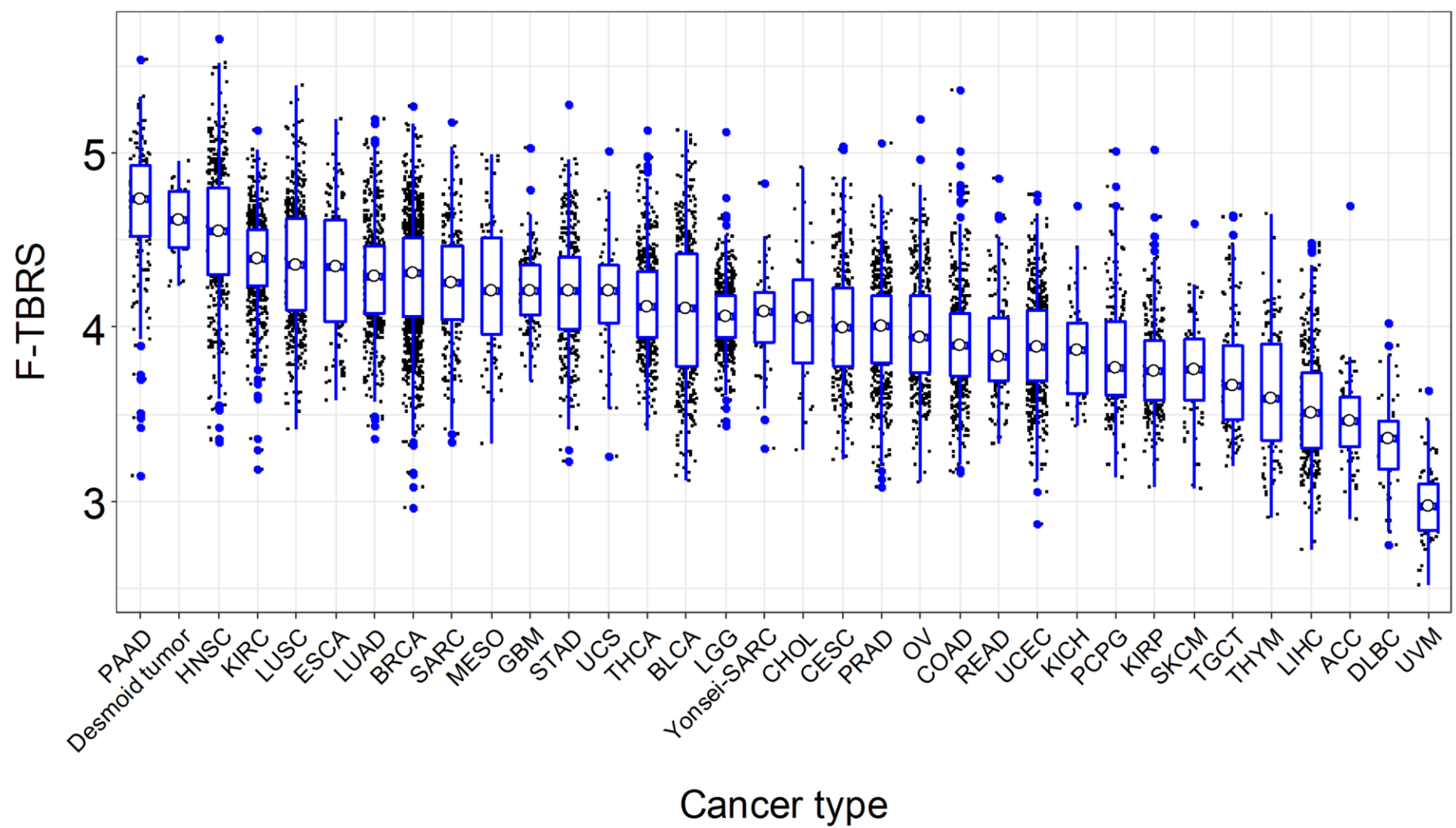
- Desmoid tumors are enriched with expression of genes associated with TGF-β response of fibroblast compared with other cancer types including other sarcomas.
- *CTNNB1*, *APC* and *GNAQ* mutations were associated with higher enrichment in TBRS.
- **Therapeutic intervention to decrease the TGF-β response of fibroblast by using TGF-β receptor inhibitor may show clinical benefit in patients with desmoid tumors.**

## REFERENCES

1. Alexandre Calon, et al., Dependency of colorectal cancer on a TGF-β-driven program in stromal cells for metastasis initiation, Cancer Cell, 2012

## ACKNOWLEDGEMENTS

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- The boxplots above show the F-TBRS levels throughout the three cohorts. Desmoid tumor samples had the second highest F-TBRS following pancreatic adenocarcinoma.