Genomic profiling reveals mutational landscape in parathyroid carcinomas

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SUPPLEMENTARY FIGURE LEGENDS

Supplementary Figure 1. Mutation distribution for the CDC73 gene in the study cohort (top) & all cancer studies in cBioPortal (bottom) along the body of the gene. The colored rectangles represent the known functional domains of the translated protein.

Supplementary Figure 2. Mutation distribution for the ADCK1 gene in the study cohort (top) & all cancer studies in cBioPortal (bottom) along the body of the gene. The colored rectangles represent the known functional domains of the translated protein.

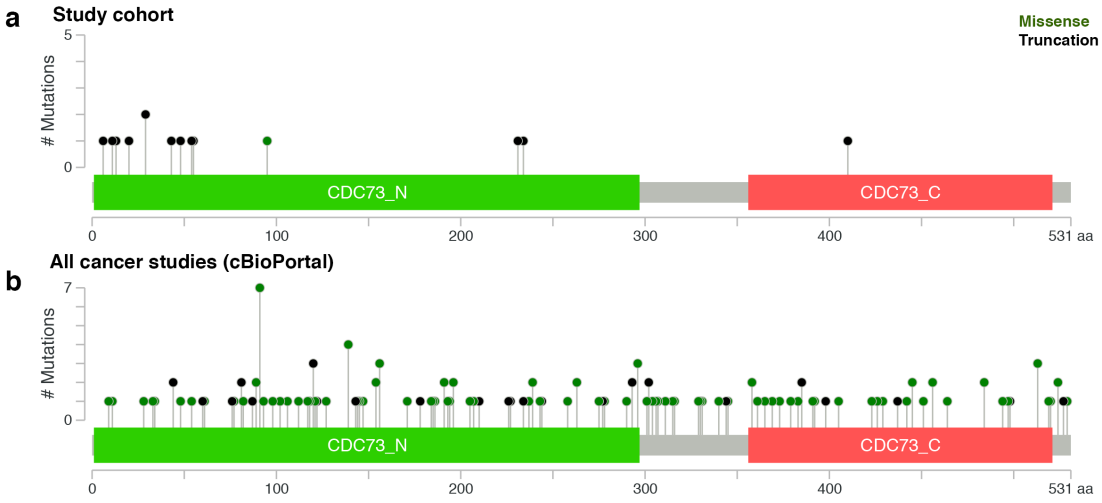
Supplementary Figure 3. Mutation distribution for the FAT3 gene in the study cohort (top) & all cancer studies in cBioPortal (bottom) along the body of the gene. The colored rectangles represent the known functional domains of the translated protein.

Supplementary Figure 4. Mutation distribution for the MTOR gene in the study cohort (top) & all cancer studies in cBioPortal (bottom) along the body of the gene. The colored rectangles represent the known functional domains of the translated protein.

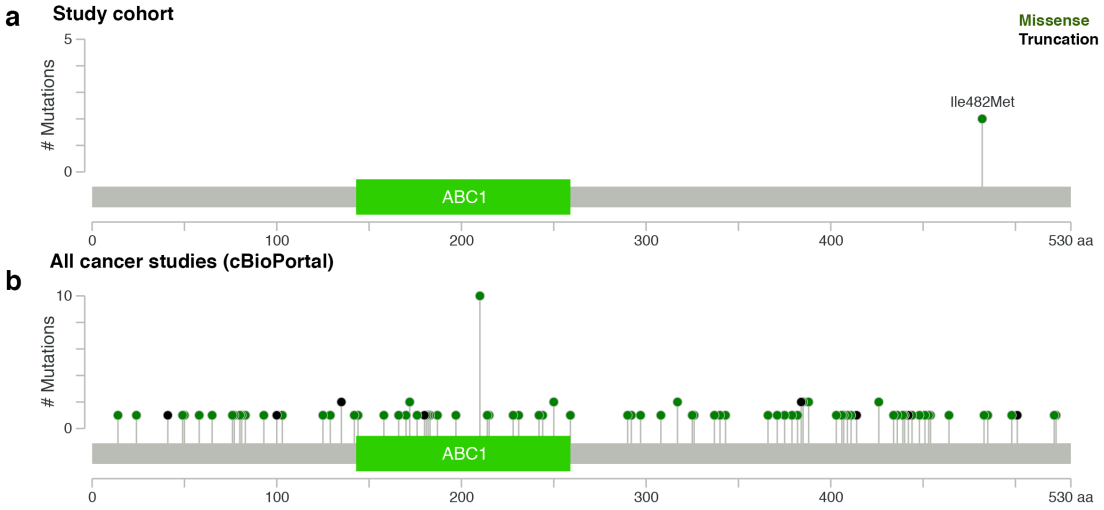
Supplementary Figure 5. Boxplots of the metrics for the ParThy panel with per-sample mean sequencing depth across all amplicons (a), total number of "PASS" variant calls (b) and variants with <2% population frequency in ESP6500, 1000 Genomes and ExAc and "high" or "moderate" predicted impact using SnpEff (c).

SUPPLEMENTARY FIGURES

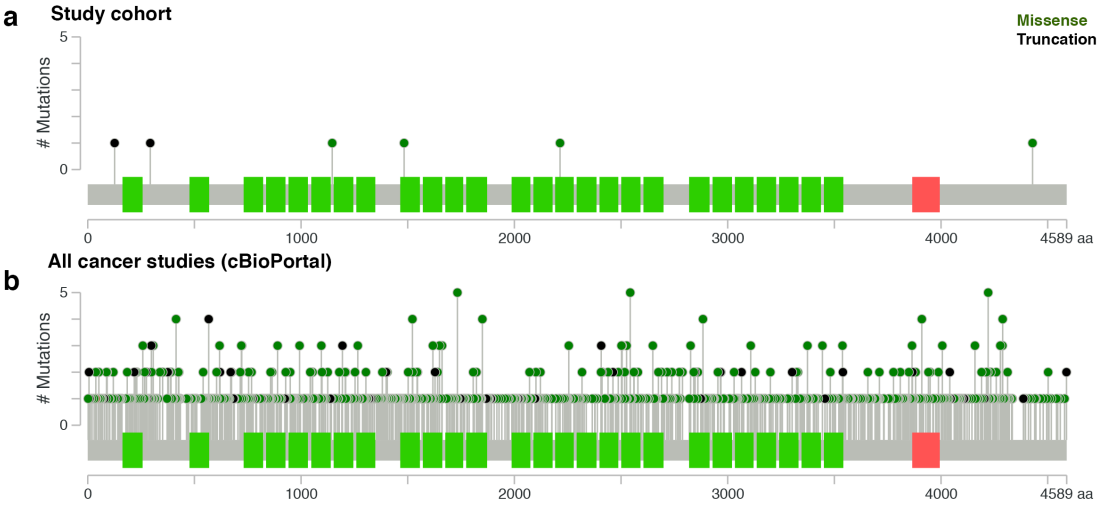
Supplementary Figure 1



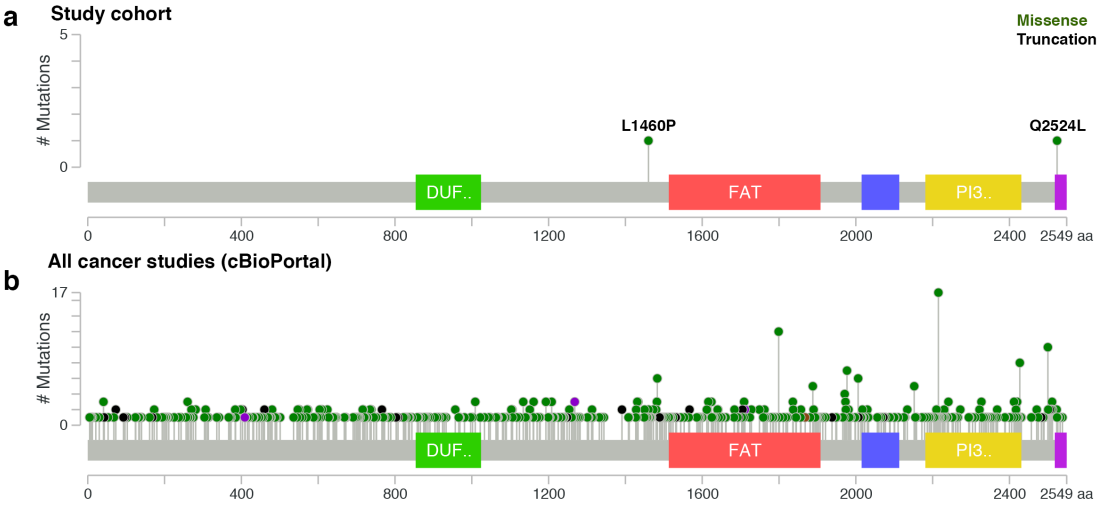
Supplementary Figure 2



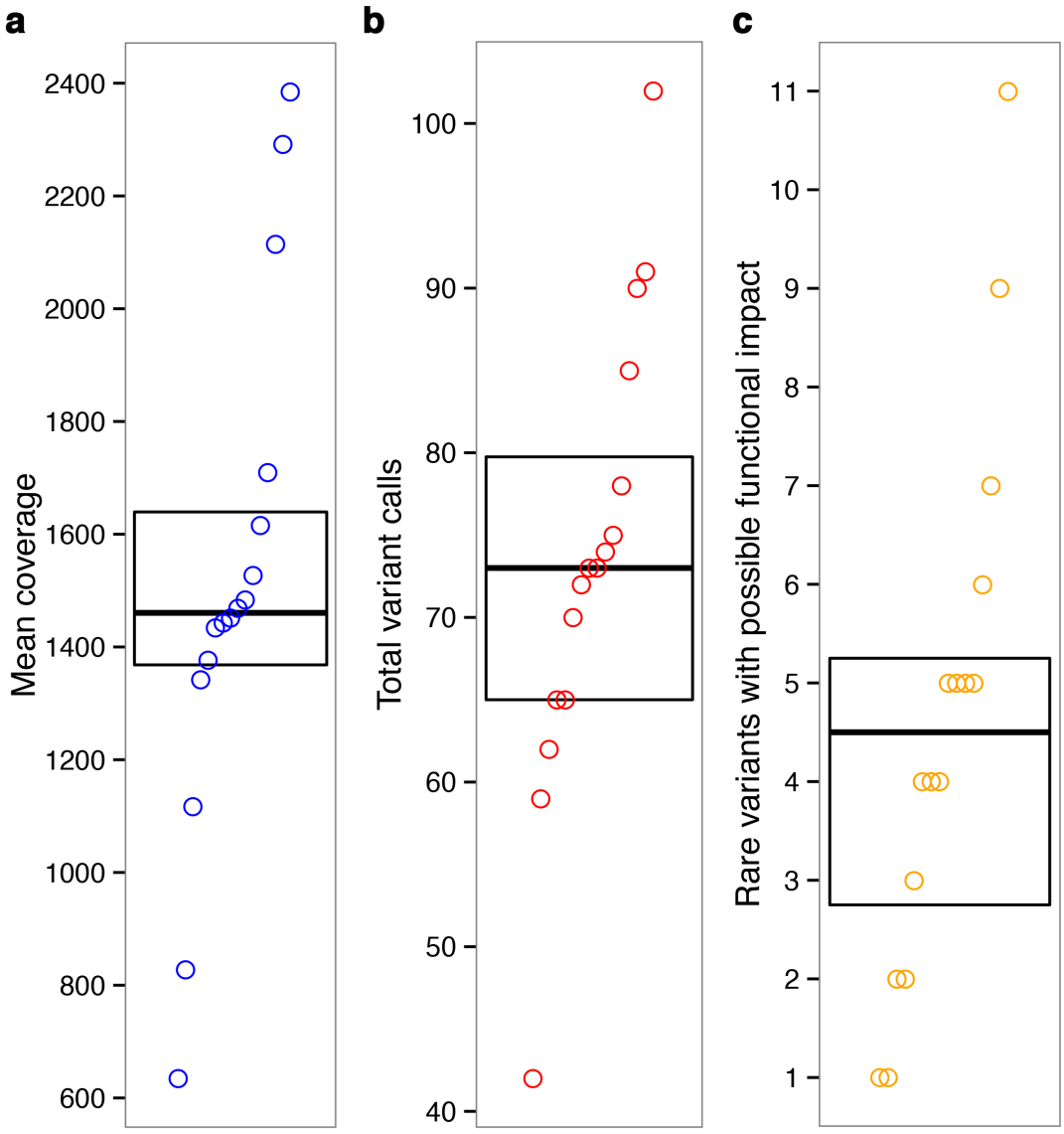
Supplementary Figure 3



Supplementary Figure 4



Supplementary Figure 5



SUPPLEMENTARY TABLES

Supplementary Table 1. Detailed QC and coverage statistics on WES data produced per sample on the discovery cohort of 17 patients.

Supplementary Table 2. Number of somatic mutations per sample.

Supplementary Table 3. List of somatic mutations in the WES discovery cohort.

Supplementary Table 4. Recurrently mutated genes in the WES discovery cohort.

Supplementary Table 5. Germline and somatic mutations in CDC73.

Supplementary Table 6. Somatic mutations in Cancer Census Genes.

Supplementary Table 7. Somatic copy number alterations as detected by saasCNV from the WES data.

Supplementary Table 8. Biallelic loss events across patients in the discovery cohort.

Supplementary Table 9. List of genes in the ParThy panel and rationale for selection.

Supplementary Table 10. Detailed QC and coverage statistics of ParThy targeted panel results for 7 tumor samples. All metrics computed using the TorrentSuite software provided by Life Techonologies.

Supplementary Table 11. List of variants detected across the 7 tumor samples using the ParThy panel.