# This Week in Cell

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A Memorial Sloan Kettering-led team tracks tumor genomic changes in individuals with advanced melanoma receiving the anti-PD-1 immune checkpoint treatment nivolumab. Using a combination of exome sequencing, transcriptome sequencing, and T-cell receptor sequencing, the researchers characterized tumor and microenvironment features before and after nivolumab treatment in 68 advanced melanoma patients, including 33 individuals who had already progressed on the CTLA-4-targeting drug ipilimumab. With these data, they were able to take a look at the tumor and immune features that corresponded to treatment response, while gaining clues to the tumor clonal selection, evolution, and immune shifts that occur following treatment.

Members of a TCGA Research Network led by investigators at the University of Texas MD Anderson Cancer Center, Brigham and Women's Hospital, and Baylor College of Medicine present a multi-omics analysis of muscle-invasive bladder cancer (MIBC). The team focused on 412 MIBC cases from the Cancer Genome Atlas project, using exome and/or genome sequencing, array-based copy number, methylation, and proteomic profiling, and RNA sequencing to identify MIBC mutation signatures, expression subtypes, and recurrently mutated genes. "Clustering by [messenger RNA], long non-coding RNA, and [microRNA] expression converged to identify subsets with differential epithelial-mesenchymal transition status, *in situ* scores, histologic features, and survival," the authors write.

The University of Washington's Evan Eichler heads a team taking a look at *de novo* mutations in hundreds of families affected by idiopathic autism spectrum disorder. Based on new or existing genome or exome sequence data for 2,064 individuals from 516 families with simplex ASD cases, the researchers narrowed in on tens of millions of single nucleotide changes and more than 9,200 private copy number variants — a collection that included nearly 134,000 *de novo* single base changes and 88 *de novo* CNVs. Relative to their unaffected siblings, they note, the ASD-affected family members were prone to *de novo* alterations that disrupted genes and produced severe mutations, including regulatory regions that are active in the fetal brain.

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bladder cancer      melanoma      MD Anderson      Memorial Sloan-Kettering      University of Washington

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