

Overview of Variant Calling Pilot 50

1000 genomes screen

- 357,458 total

Sensitive

- Sensitive: 933
- Ultra-Conserved: 23,013 GERP>3.0

Recurrence

- TTN (chr2:179390716-179695529) Altered in 11/41(26.83%) samples.
- TP53 (17p13.1) Altered in 8/41(19.51%) samples.
- Enhancer(drmlchr6:28739200-28741300) Altered in 5/41(12.20%) samples.

Cancer implicated

- 12,535