



ABSTRACT

“SHOREmap: simultaneous mapping and mutation identification by deep sequencing”

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Identification of causative point mutations after forward genetic mutant screens typically begins with genetic mapping, followed by transformation rescue or candidate gene sequencing. We present a one step alternative: performing hundreds of thousands of genotyping assays while sequencing all candidate genes. This is accomplished by deep sequencing of a pool of F2 progeny obtained by crossing to a polymorphic strain and does not require prior knowledge of mapping markers. The application of high-throughput sequencing shortens the overall time required for genetic mapping from months to weeks and, importantly, greatly reduces investigator hands-on time. The steps requiring investigator input are: DNA isolation (1 day), library preparation and validation (4 days), Illumina cluster generation and sequencing (2 days), and data analysis (1 day). Once the mapping population has been established, the present method therefore allows a single investigator to identify a causative mutation within only eight working days – approximately an order of magnitude faster than with conventional methods.