

BUDGET JUSTIFICATION – The Jackson Laboratory for Genomic Medicine

SENIOR/KEY PERSONNEL

Charles Lee, Ph.D., Principal Investigator (Contact PI) (1.2 calendar months), is Scientific Director and Professor at The Jackson Laboratory for Genomic Medicine (JAX-GM) will serve as Principal Investigator and Contact PI. As Contact PI, he will coordinate communication among the PIs, key personnel, and NIH scientific and program officials, and will be responsible for coordinating the preparation and submission of annual progress reports. Dr. Lee has over 15 years experience successfully leading both R01- and collaborative U-mechanism projects, with a particular emphasis on the detection of genetic variants using state-of-the-art technologies. For this project, he will be responsible for the overall scientific direction of the development of an integrative pipeline of tools for complex structural variation (SV) discovery and *in silico* validation of SV events discovered in the analysis of genome sequence datasets being generated by the various TOPMed projects.

Ankit Malhotra, Ph.D., Co-Investigator (3.0 calendar months year 1, 2.4 calendar months years 2-3) and Associate Computational Scientist at The Jackson Laboratory, has extensive expertise in genome analysis, especially in the field of structural variations (SV) in the human genome. As a postdoc, he was recipient of a prestigious Department of Defense (DoD) Breast Cancer Research Program (BCRP) fellowship for his work on characterizing structural variations in breast cancer patients. He also lead one of the first meta analysis of complex structural variations in patient datasets from The Cancer Genome Atlas (TCGA) consortium and has developed tools to discover complex SV from whole genome sequencing datasets. He will be involved in novel method development and analysis of sequencing datasets from the different centers producing the genome sequence datasets being generated by the various TOPMed projects. Working together with Dr. Lee, he would also be responsible for mentoring the two postdocs being funded for their work on this project.

OTHER PERSONNEL

Support is requested for To-Be-Named project-driven postdoctoral level **Computational Scientist(s) (12 calendar months total per year)**. The postdoc(s) will direct effort towards developing the methods for discovery and *in-silico* validation of the whole genome sequencing datasets. They will also be involved in the meta analysis of population structure and recombination rates using the identified SVs and the impact of identified on disease biology.

FRINGE BENEFITS

Fringe benefit costs have been calculated at 30% of salary in accordance with the Rate agreement negotiated between The Jackson Laboratory and DHHS at the time of proposal submission.

TRAVEL

\$3,000 per/year is requested to accommodate one trip for each of the key investigators at JAX-GM to attend and participate in an in-person meeting with NIH U01 TOPMed Project representatives. The estimated cost of \$750 for each key individuals includes coach airfare (\$250) to Washington, one night lodging (\$250) and two days per diem expenses and ground transportation (\$250). In addition, the remaining \$1,500 is requested to support the costs of two researchers to attend and participate in an annual scientific meeting.

OTHER EXPENSES

Materials and Supplies

Lab supplies Funds are requested for office supplies, disposables, and other materials needed for the in house computing work taking place at Jax-GM.

SUBAWARDS/CONSORTIUM/CONTRACTUAL COSTS

A letter of intent to enter into a formal written consortium agreement for meeting the scientific, administrative, financial, and reporting requirements of the proposed project has been executed with both Yale University and the McDonnell Genome Institute at Washington University in St. Louis. The proposed total costs for Yale are \$663,248 and the proposed total costs for the McDonnell Genome Institute is \$608,475.

FACILITIES AND ADMINISTRATIVE COSTS

The Facilities and Administrative costs have been calculated at 89% of MTDC for the Farmington, CT campus in accordance with the Rate Agreement negotiated between The Jackson Laboratory and DHHS at the time of proposal submission.