

Survey on bioinformatics resources for human genome variation

Dear life science professional,

we are contacting you to kindly ask if you could help us by using 10-15 minutes to participate in a survey on bioinformatics resources for human genome variation. The survey aims to understand and improve the usage of existing infrastructure, resources and protocols for human genome variation annotation and curation. The focus is on processes that can be automated to support interpretation of high-throughput genome sequencing results for research as well as for clinical use.

You can fill out the survey here: <https://www.webropolsurveys.com/S/AEB30ED3EDBE6121.par>

The outcome of the survey will be a report that describes the current status within ELIXIR member states, identified requirements and potential solutions. The report will be part of the ELIXIR Human Genomics and Translational Data Services strategy and roadmap, and will be published on the ELIXIR website in late 2019 (<https://www.elixir-europe.org>).

ELIXIR is a non-profit, intergovernmental organisation that brings together life science resources from across Europe. These resources include databases, software tools, training materials, cloud storage and supercomputers. The goal of ELIXIR is to coordinate these resources so that they form a single infrastructure. This infrastructure makes it easier for scientists to find and share data, exchange expertise, and agree on best practices. Ultimately, it will help them gain new insights into how living organisms work.

The survey is open until September 30th, 2018. Please note that the answers will only be sent after pushing the “Submit” button. Your answers will not be saved if you quit before that, but you can start over by opening the link again. The survey is anonymous.

If you have questions related to the survey, please contact Mats Lindstedt, CSC - IT Center for Science (mats.lindstedt@csc.fi) or to myself (marcella.attimonelli@uniba.it).

Thank you for your help.

Sincerely,

Marcella Attimonelli

Elixir-IIB, The Italian Elixir Node – Unit of University “Aldo Moro”, Bari (IT)

On behalf of the ELIXIR team for the Implementation study on annotating and curating human genomic variants

The survey is run by the following ELIXIR Nodes and partners:

CSC - IT Center for Science Ltd., Finland

National Institute for Health and Welfare (THL), Finland

Mitochondrial Bioinformatics Unit - University of Bari, Italy

SIB Swiss Institute of Bioinformatics, Switzerland

The European Bioinformatics Institute (EMBL-EBI), UK

Imperial College London, UK