

## Bioinformatics Support Request

Please provide us with more information on your request for support. Complete the form as comprehensively as possible, and please indicate where there is still uncertainty.

**Please note, the earlier we are involved the better – for example, it would be better for us to be involved during the study design and even grant application stage.**

CONTACT DETAILS	
Date of request	02/03/2022
Name	Sune Mostert
Email address	<a href="mailto:MSTSUN003@myuct.ac.za">MSTSUN003@myuct.ac.za</a>
Research Group/Department	Forensic Pathology and Toxilocology
Faculty	Health Sciences
IF student, name & email of supervisor	Dr Laura Heathfield, laura.heathfield@uct.ac.za

PROJECT DETAILS
1. What is the scientific question?
We will be analyzing next generation sequencing data to perform variants calling in order to identify a possible genetic variant that could contribute/better explain the sudden unexpected death of an infant (SUDI) (less than 1 year old).
2. Who are the partners on the project?
No partners. This study will be investigating a case form Salt River Mortuary.

3. What type of collaboration with CBIO is expected? For a project that is done as collaboration or for a fee, we will put the agreement in writing.
A consultation with one of the bioinformaticians is expected, providing insight in the appropriate software to use, recommendation of training in order to execute the pipeline with all the necessary quality control checks in between. I will be performing the bioinformatic analysis myself.
4. Are there any ethical issues we should be aware of?
This data is medico-legal of nature and only authorized personnel will have access to the data.
5. How much work is expected from CBIO and when?
The input expected is mostly that of giving advice and direction.
6. What type of data will be generated (e.g. sequencing, genotyping, expression, etc.) and what technology platform will be used?
Fastq files was generated from Next generation sequencing.
7. When do you expect the data? Does it need to be transferred from somewhere else?
The sequencing has already been performed. I am awaiting ethical approval in order to start the study which will be analyzing the data that has previously been generated.
8. How large will the data be? How long does it need to be stored for, and have you made arrangements for storage?
The total size of the data is 32.8 GB. The data can be stored for as long as the pathologist

requires it. Arrangement for the storage has been made.

**9. What bioinformatics analysis needs to be done? Which tools are required?**

Human variant calling has to be performed.

**10. If a collaborative model is being used, what papers are envisaged and who will the authors be?**

**11. Can we add a short description and objective of the project to the CBIO website?**

Yes, however the medico-legal nature of the project will have to be taken in consideration when writing the description of the project.

**PLEASE FORWARD THE COMPLETED FORM TO:**

[Nicola.mulder@uct.ac.za](mailto:Nicola.mulder@uct.ac.za)