Institut für Biomedical Informatics

Master thesis (MA, 30 ECTS)

Identification of structural variants in the cancer genome

Description

Structural variations (SVs), including inversions, deletions, duplications, and translocations, are a hallmark of most cancer genomes. The discovery of recurrent SVs and their consequences for genome organization and gene expression has greatly advanced our knowledge of oncogenesis. Nevertheless, identifying SVs in cancer genomes remains challenging.
We are collaborating with the groups of PD Dr. Hugo Murúa-Escobar (Universitätsmedizin Rostock) and Prof. Ingo Nolte (Stiftung Tierärztliche Hochschule Hannover Klinik für Kleintiere) and have access to a number of whole-genome sequencing (WGS) and RNA-seq libraries of canine prostate and bladder cancer samples as well as derived cell lines. The aim of this project is to develop a framework integrating WGS and RNA-seq to detect SVs and their effects.

Relevant literature

- Cameron DL, Di Stefano L, Papenfuss AT. Comprehensive evaluation and characterisation of short read general-purpose structural variant calling software. Nat Commun. 2019 Jul 19;10(1):3240. doi: 10.1038/s41467-019-11146-4.
- Eisfeldt J, Pettersson M, Vezzi F, Wincent J, Käller M, Gruselius J, Nilsson D, Syk Lundberg E, Carvalho CMB, Lindstrand A. Comprehensive structural variation genome map of individuals carrying complex chromosomal rearrangements. PLoS Genet. 2019 Feb 8;15(2):e1007858. doi: 10.1371/journal.pgen.1007858.

Requirements

Familiarity with UNIX, bash, a scripting language (Perl or Python), and R. These skills could be acquired during the preparation of the thesis, but this would require substantial additional time and effort.

Start date

As soon as possible

Duration

Approximately six months

Contact

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