

Position for Tertiary NGS data bioanalyst (m/f)

Internal code: CGPP2001

The Centro de Genética Preditiva e Preventiva (CGPP, www.cgpp.eu), the unit for provision of medical genetics services of the Institute for Molecular and Cell Biology (IBMC, www.ibmc.up.pt), at i3S (www.i3s.up.pt), is opening a position for a tertiary data bioanalyst of next-generation sequencing (NGS) data, for clinical molecular genetics testing. The successful candidate will engage in NGS data analysis, for the identification and classification of disease-causing variants in hereditary diseases, using cutting-edge genomic technologies.

Minimum prerequisites: The candidate must have (1) a background in Biology, Biochemistry or related fields in the area of the life or health sciences; (2) good background knowledge about human and medical genetics; (3) previous experience with molecular biology techniques; (4) expertise on tertiary data analysis of NGS results; (5) experience using a variety of software and databases, in particular for classification and interpretation of sequence variants; (6) knowledge about quality procedures at a molecular genetics laboratory; and (7) strong teamwork spirit and good communication skills.

Preferential conditions: (1) MSc or PhD in human molecular genetics or related fields; (2) Registered Clinical Laboratory Geneticist by the European Board of Medical Genetics (EBMG) or at least three-year experience working at a clinical molecular genetics laboratory; (3) expertise in NGS data analysis and interpretation in the context of clinical genetic testing; (4) familiarity with the following software (in the perspective of an user): Alamut, IGV and SeqScape and with genetic diseases/variants databases; (5) previous experience in NGS data processing in an Unix environment, namely proficiency using Linux command line including shell scripts; (6) the clinical laboratory knowledge and expertise necessary for generating medical diagnostic reports; and (7) acquaintance with quality management systems for (accreditation of) clinical laboratories (ISO 15189).

Applications are open from July 1st to July 20th, 2020. The expected starting date would be 1th September 2020.

The proposals must include a letter of motivation, a short CV, copies of degrees' diplomas and a minimum of one reference contact (or reference letter) and must be submitted at the link:

<http://www.ibmc.up.pt/gestaocandidaturas/index.php?codigo=CGPP2001>

Jury: President – Jorge Sequeiros (MD, PhD); Ordinary members – Jorge Oliveira (PhD), João Parente Freixo (MD), Milena Paneque (PhD) and Paulo Silva (PhD)