

NECESSITY REPORT TO ESTABLISH THE HIRING THE SOFTWARE LICENSES FOR VISUALIZATION, INTERPRETATION, AND IN SILICO PREDICTOR SOFTWARE FOR THE GENETIC MEDICINE RESEARCH GROUP OF THE FUNDACIÓ HOSPITAL UNIVERSITARI VALL HEBRON – INSTITUT DE RECERCA (VHIR)

NEG 2023-021 SOFTWARE ALAMUT VISUAL PLUS

Dra. Elena García Arumí, of the Genetic Medicine Research Group of the Fundació Hospital Universitari Vall d'Hebron - Institut de Recerca, has proposed a procedure for the **“HIRING THE SOFTWARE LICENSES FOR VISUALIZATION, INTERPRETATION, AND IN SILICO PREDICTOR SOFTWARE FOR THE GENETIC MEDICINE RESEARCH GROUP OF THE FUNDACIÓ HOSPITAL UNIVERSITARI VALL HEBRON – INSTITUT DE RECERCA (VHIR)”**.

The molecular characterization of genetic variants, with the aim of identifying potentially pathogenic variants and identifying the consequences of genetic diseases, represents one of the indispensable research activities of the Genetic Medicine Research Group. To carry out this task, the annotation, interpretation and visualization of genetic variants is essential. Alamut Visual Plus software is the only software on the market that offers bioinformatics databases and predictors of pathogenicity, both splicing and protein in a unique way. In addition to the integrative visualization of the various databases, as well as the versatility of bioinformatics files with which it can manage.

The division of the subcontract into lots is not considered advisable, since the award to a single bidder will facilitate the proper execution of the service from a technical and organizational point of view and organizational point of view. The division into lots would make the execution of the contract excessively difficult or onerous from a technical or organizational point of view technically difficult or onerous, or that the need to coordinate the different contractors for the various lots could seriously risk undermining the proper execution of the contract.

The duration of this purchase contract will be of two (2) years. The initiation of the contract shall be on the day following the date of formalization of the contract.

The maximum approved budget for the tender for the provision of the services for to the total duration of the procedure, is “NINE THOUSAND SIX HUNDRED AND NINETY-SIX EUROS” (€ 9.696,00), VAT exempted.

The annual unit budget for each licence is “TWO THOUSAND FOUR HUNDRED AND TWENTY-FOUR EUROS” (€ 2.424,00), VAT excluded.

The contracting entity will not be obliged to exhaust the entire base budget for the tender, but will only be billed for the services actually executed based on the prices offered by the contractor.

In view of the need to carry out a negotiated procedure without advertising for reasons of exclusivity, and non-harmonized regulation according to criteria established in Law 9/2017, of 8 November, on Public Sector Contracting, whereby Directives of the European Parliament and of the Council 2014/23/EU and 2014/24/EU, of 26 February 2014, are transposed into Spanish law.

In view of all of the above,

IT IS HEREBY DECIDED:

- 1.- To order the opening of the tender file for the **“HIRING THE SOFTWARE LICENSES FOR VISUALIZATION, INTERPRETATION, AND IN SILICO PREDICTOR SOFTWARE FOR THE GENETIC MEDICINE RESEARCH GROUP OF THE FUNDACIÓ HOSPITAL UNIVERSITARI VALL HEBRON – INSTITUT DE RECERCA (VHIR)”**. The maximum budget for this tender is set at 9.696,00€ VAT exempted.
- 2.- To order the certification of the existence of sufficient credit to certify that the VHIR has a sufficient budgetary endowment to face the expenses arising from this tender file.
- 3.- To draft the Specific Administrative Bidding Conditions and Technical Specifications in accordance with the provisions of the aforementioned Law.

Barcelona, on the digital signature date.

RESPONSIBLE

Dra. Elena García Arumí

Genetic Medicine Research Group

Fundació Hospital Universitari Vall Hebron - Institut de Recerca (VHIR)

Hospital Universitari Vall d'Hebron
Pg. de la Vall d'Hebron, 119,
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Spain

CERTIFICATE OF EXCLUSIVITY

ALAMUT VISUAL Plus

Alamut Visual Plus is an exploration software application for genomic variations that integrates genetic and genomic information from different sources into one consistent and convenient environment. It gathers in one place a wide set of external data and algorithms of recognized quality that are useful to the biologist for the interpretation of genomic variants.

To the best of our knowledge, the product commercialized by SOPHiA GENETICS offers a unique combination of features (stated hereunder), and there are no other software package offerings, through other companies or entities, offering this combination of features.

The unique combination of features of Alamut Visual Plus is the following:

- Graphical and interactive genome browser including high-quality public genome annotations (see Alamut Database below);
- Display of genes and transcripts reference sequence data from RefSeq, HUGO, Ensembl;
- Investigation of several genes at one time
- Integration of the HGVS (Human Genome Variation Society) recommendations for the description of sequence variants;
- Integration of missense variant effects and of RNA splicing effects prediction algorithms;
- Generation of customizable variant reports including potential pathogenicity classification from external data sources;
- Display of standard bioinformatic files (BAM/CRAM, VCF,BED) ;
- Display of Sanger electropherogram
- Management and display of private imported variants locally and storage on user's computer or on a local network;
- Management and display of locally stored sequence annotations.

Alamut Database

This database provides a collection of data that are relevant for human genome variant annotation and interpretation.

The Alamut Database integrates data from the following sources:

- Human reference assembly (GRCh37 et GRCh38)
- Human Mitochondrial reference sequence (rCRS)
- Genomic sequences LRG
- Human Genome Organisation (HUGO) HGNC;
- Transcripts: NCBI RefSeq, Ensembl, MANE transcripts
- OMIM® database

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- dbSNP, ClinVar
- GoNL, HGVD, Danish2K
- UCSC Genome Conservation scores (PhyloP and phastCons)
- EMBL-EBI Ensembl, UniProt, InterPro, Orthologues;
- Broad Institute gnomAD;
- 1000 Genome;
- NHLBI GO ESP;
- Sanger Institute COSMIC;
- HPO DGV and RepeatMasker
- Abstarct from PubMed
- Mastermind
- Splicing predictors (SSF-Like, MaxEntScan, Genesplicer, NNSPLICE)
- Missense predictors (AlignGVGD, SIFT, MutationTaster)
- Linkouts to external databses (LSDB, OMIM, NCBI, GENEATLAS, UCSC)

We, the undersigned Mr. Ken Freedman and Mr. Esteban Czwan acting on behalf of the company, certify the exclusivity of the service offered to Hospital Universitari Vall d'Hebron.

Product concerned by this certificate:

- ALAMUT Visual Plus - Tier B - 2 tokens - ALA AVA-B

Hospital Universitari Vall d'Hebron acknowledges that the referenced product is not approved as an *in vitro* diagnostic product in Europe. Accordingly, Hospital Universitari Vall d'Hebron accepts sole and exclusive responsibility for the interpretation of any results and data provided by SOPHiA GENETICS and the provision of clinical genetic diagnosis on the basis of said results and data.

Rolle, Switzerland

23.05. 2023

Legal Representative

DocuSigned by:
A blue ink signature of Ken Freedman.
D06056FBA833451...

M. Ken Freedman
SVP, Chief Revenue Officer

DocuSigned by:
A blue ink signature of Esteban Czwan.
3B666E10038A40C...

M. Esteban Czwan
Managing Director EMEA