

FOURTH Pirepred MEETING

18 January 2018

Aula 4, planta 11. Àrea General Hospital Universitari Vall d'Hebron

16:00 h. Gathering and Welcome (by Xavier de la Cruz).

16:15 h. Announcements (by Javier Sancho).

16:30 h. State of Pirepred Action n. 4

ACTIVITY 4.1 Analysis of patient mutations

State of flow of mutations within Pirepred (by Xavier de la Cruz and all others).

Progress in variant reporting in PIREPRED (by Juan Fernández-Recio).

State of deliverables (by Javier Sancho).

- Individual reports for interpreted mutations: description of mutation, clinical associated data, bioinformatic evaluation, date of order, delivery and delivery to hospital of origin.
- Annual document that gathers all the reports: **to deliver at next meeting.**

ACTIVITY 4.2. Development of prediction and evaluation methods

Progress in prediction methods (by Juan Fernández Recio and Javier Sancho).

State of deliverables (by Javier Sancho).

- Internal document about software development strategy.
- Document that describes a set of mutation tests with different objective degrees of difficulty.
- Internal documents that describe the results of the different approaches implemented to improve the function of pathogenicity score and optimal function.

17:45-18:00 h. Break

18:00 h. State of Pirepred Actions n. 1, 2 and 3

ACTIVITY 1.2. Follow up of project (Javier Sancho).

State of declarations/certifications.

Follow up meetings: dates and locations.

ACTIVITY 2.2. Diffusion actions (all partners).

Report on diffusion activities, and justification material.

Final scientific meeting in Zaragoza (2019/08/31): committees, dates and ideas.

Proposals for networking with relevant patient's associations.

ACTIVITY 3.2. Cooperation activities of the Pirepred Network (all partners and associated partners).

Pirepred new member proposal.

18:30 h. Summary of the tasks to be carried out

18:45 h. Closing