

### **THIRD Pirepred MEETING**

6 July 2017.

#### **Sala Sarasate**

BioCruces .Hospital Universitario Cruces.  
Plaza de Cruces 12, 48903. Barakaldo, Bizkaia (País Vasco)

**9:00 h. Gathering and Welcome** (by Koldo Aldámiz-Echevarría)

**9:15 h. Announcements** (by Javier Sancho)

**9:30 h. Tasks from Pirepred second meeting.**

Deliverables (15 min each one).

- Deliverable: document Neonatal screening programs in France (*Frédérique Savagner*)
- Deliverable: document with mutations detected in this period (*Inmaculada García, Koldo Aldámiz- Echevarría, and Felix Sánchez from Spain; Frédérique Savagner and Isabelle Olivier from France*)
- Deliverable: document on mutation analysis by Molecular Dynamic simulation (*Javier Sancho*)
- Deliverable: document on phenotype prediction methods (*Juan Fernández Recio*)

**10:30 h. State of Pirepred specific actions**

**10:30 h. Pirepred network development ACTION 3**

**Network cooperation activities A3.2**

- Brief exposition of the state of neonatal screening programs in France (*by Frédérique Savagner*)
- Pirepred new member proposal (*by all partners and associated partners*)

**11:00 h. Coffee break**

**11:30. Analysis of mutations and improvement of interpretation methods ACTION 4**

**Analysis of patient mutations A4.1**

- Presentation of current flow of mutations within Pirepred (*by Inmaculada García and Koldo Aldámiz- Echevarría*)
- Progress in variant annotation and reporting in PIREPRED (*by Xavier de la Cruz*)

**Development of prediction and evaluation methods A4.2**

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

**13:00 h. State of Pirepred general actions**

**12:30 h. Pirepred management ACTION 1**

**Administrative and financial follow-up (coordinator partner) A1.2**

- Follow meetings: date and location

**13:00 h. Pirepred communication ACTION 2**

**Dissemination actions A2.2**

- Scientific meetings organized (enumeration and brief description by partners)
- Proposals for focused brief meetings/activities
- Proposals for networking with relevant patients associations

**14:00 h. Summary of the tasks to be carried out**

**14:10 h. Closing**

**14:15 h. Lunch break**