

16 January 2016.

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Centre National de la Recherche Scientifique UMR 5089 DR14
205 route de Narbonne, 31077 Toulouse, France*

9:00 h. Gathering and Welcome (by Olivier Cuvillier)

9:15 h. Announcements (by Javier Sancho)

9:30 h. State of Pirepred specific actions

9:30 h. Pirepred network development ACTION 3

Network cooperation activities A3.2

- Scientific meetings organized (*enumeration by partners*)
- Brief exposition of the state of neonatal screening programs in the different regions (*by Inmaculada García and other members*)

10:00 h. Analysis of mutations and improvement of interpretation methods ACTION 4

Analysis of patient mutations A4.1

- Brief description of the current flow of mutations within Pirepred (*by Inmaculada García*)
- An initial draft of a mutation report (*by Xavier de la Cruz*)

Development of prediction and evaluation methods A4.2

- Brief description of ongoing and planned activities (*by Juan Fernández*)

10:30 h Parallel working sessions

Clinicians (partners and associates) meet and discuss on:

- Possibilities and proposals on new associate members in not represented regions
- Organization/beginning of the drafting of document on the present of neonatal screening in the different regions and of a consensus recommendation report (extension, prioritization, resources) for health authorities
- Organization/beginning of the drafting of document for patients associations

Bioinformatics meet and discuss on:

- Organization/beginning of the drafting of document describing Pirepred software development strategy
- Proposals for a specific test set of mutations

12:00 h Join Discussion

12:30 h. Lunch break

14:00 h. State of Pirepred general actions

14:00 h. Pirepred management ACTION 1

Administrative and financial follow-up (*Yolanda Vergara*) A1.2

14:30 h. Pirepred communication ACTION 2

Dissemination actions A2.2

- Proposals for focused brief meetings/activities
- Proposals for networking with relevant patients associations

15:00 h. Summary of the tasks to be carried out

16:00 h. Closing