

1ST EUROPEAN
DAYS OF
ALBINISM

Organized by
association française
Genespoir
des albinismes

October 27th & 28th 2012
Paris, FRANCE

**Report of participation in the first European
conference albinism**



On October 27th and 28th, "First European days of albinism" the first European conference ever dedicated to albinism was held in Paris (France). Italy was represented by [Albinit](#) and [Albinismo.eu](#): they have had the great opportunity to present themselves, drawing an outline of albinism in Italy and illustrating current projects and initiatives. Giancarlo Loddo (from [Albinismo.eu](#)), in particular, proposed an interesting video, showing the important contributions of his free and independent community. [Albinit](#), on the other hand, projected a short PowerPoint proving, more than any other word, the potential and limitations of still a young and structuring association.

The meeting's main purpose consisted, on one hand, to share experience between members demonstrating the everyone's limits and potential, on the other, to promote dialogue between researchers, patients and family members of people with albinism in order to illustrate researching performance and define a road map for the future

The program was structured in two sessions. In the first (on Saturday 27th) associations and researchers met themselves separately to identify common needs and expectations for the future. The result was a statement of intents (that will be improved and confirmed in the coming weeks), so I'm going to describe it shortly. On Sunday instead researchers (including Dr. Patrosso from



[Niguarda](#) Hospital in Milan), showed to the associations their commitments focusing on the future perspectives in research. Considering the participant realities (from Italy, Spain, France, UK, Germany and USA) it is possible to draw some preliminary conclusions.

1. Structure, membership and operating tools

Generally French, British and American associations appear highly structured, with a massive number of members (ranging from about 400 of Genespoir to thousands of NOAH, a formal structure and a robust and developed multimedia channel (website, forums, facebook groups etc.). [Albinit](#) and Alba (the Spanish association) collected respectively 60 and 130 members, are less structured and the relationship among members is based on more casual and light communicative register. [Albinismo.eu](#), on the other hand, is characterized by completeness and thoroughness of the information provided and vital contribution of the three national meetings occurred between 2009 and 2012

Here you are a couple of pictures providing a short profile of the European associations and number of members.



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As you can see, the most recent associations get limited number of members; in total there are 1500 members in respect of all associations, a very small number compared to the geographical coverage.

Apparent cultural differences between Anglo-Saxon and Mediterranean world, however, should not be misunderstood. In Italy there are about 3000-3500 people with albinism, so the membership in a community or association is not going to achieve particularly high percentages in the near future. Furthermore in our culture (especially in Italy) persists a more defeatist attitude, fear to get involved, and perpetual worry of encountering prejudice and discrimination

2. Common difficulties

Despite the strong structure and long life of the Anglo-Saxon colleagues, difficulties remain common within all realities considered, including:

- Difficulty to encourage research
- Fund raising problems (apart NOAH that receives governative funding)
- Widespread ignorance among the scientific community and the general public about the issues that albinism involves.
- Extreme heterogeneity of legislation on disability in every represented country



- Need to raise the public awareness through accurate and responsible information campaigns
- Coordination Difficulties
- Frequent leak of the members
- Passive parent `s attitude

3. Italian recognition

A great merit recognized by all scientists to the Italian community (the research community, [Albinit](#) and [Albinismo.eu](#)) was the contribute to the creation of a [multidisciplinary diagnostic path](#) (at the [Niguarda](#) Hospital) that has no equal in other European countries. The contribution of Maria Cristina Patrosso and all researchers from [Niguarda](#) was greatly appreciated by the entire international community and is in many countries a reference to look at. The hope in the future will be to replicate similar initiatives in their own County.

4. Scientific contributions

The albinism research is getting an hard time in both Europe and USA, for the limited amount of available resources (and troubles to find new ones). The researcher's tendency to follow old paths contributes too. Therefore the genetic mapping at [Niguarda](#) assumes great importance in the perspective of a therapeutic approach focused on each single identified mutation. Currently scientists are studying the effects of certain drugs



(including Nitisinone) that would be able (in OCA1B patients) to stimulate tyrosinase and thereby increase the production of melanin (with consequent benefits for the skin and eyes). These are substances whose benefits on patients with albinism have long been known: at now however, there are no human experimentation able to show real benefits from that new drugs .

5. Proposals

The Associations agreed on the following proposals (that will result in a declaration of intent for the future)

- The establishment of a shared virtual space in which the main initiatives of each association / group / community (like a sort of portal or bulletin) will be reported.
- The need to preserve, despite the sharing of ideas and goals, single association's identities (many issues are specific to each European country)
- The common need to exert strong pressure on the scientific community to raise awareness about the challenges that albinism involves
- The importance to involve at the same level people with albinism and their families, accompanying the children's growth
- Meeting's promotion (both formal and informal)
- The network development to actively involve near and far people



- Communication campaigns based on balance, responsibility and wisdom (avoiding the risk of a sterile victimization)
- Dialogue enhancement between patient and doctors to inform and get informed about the visual and dermatological issues.
- Strong pressure against pharmaceutical lobbies to put albinism at the center of their agenda, by a "persuasive" approach: new active strategies could be useful not only to albinism research, but also to many other rare diseases.
- The importance to join European organizations like Eurordis working to give voice within the institutions to patients with rare diseases and their associations, facilitating project development and campaigns

From the scientific community emerged the following considerations:

- The key contribution of associations in orienting research towards clear objectives, defining the priorities (low vision, skin?)
- Considering the wide range of events and issues that albinism involves (visual acuity ranging from 1/20 to 5-6 tenths, varying hypopigmentation, et.) specific therapy oriented on each person and his characteristics could be useful



6. Conclusions

The meeting emerged like a positive experience. It allowed participants to discuss, understanding the single association's strengths and weaknesses. Language barrier's overcoming is a major problem: English is essential to be a world citizen. Italy get immense potential, but often it doesn't show it . I think we succeeded to illustrate the life of our Italian realities their limitations and potential. Respecting the [Albinit](#) and [Albinismo.eu](#) structural characteristics and identity, the need for a collaborative and synergistic approach is getting more and more apparent and will be fundamental in the near future. In this sense, regardless of the specificity of both, there are plenty of scope for future collaboration, useful to avoid waste of energy and time: that could facilitate as patients (people with albinism), as doctors (scientists, ophthalmologists, dermatologists etc.). In fact new strategies, diagnostic pathways, as the attempt to amend the current, incomplete legislation on disability should be more approached as a joint effort. The statement of intents that will emerge from the experience in Paris, finally, is going to take the form of a balanced report, more focused on everyone's expectations, than on specific characteristics and difficulties.

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