The “Global Naevus Initiative” case study

Stefan Wilms¹, Heather Etchevers², ³ and Mark Beckwith⁴

¹Nevus Network Nederland, Enschede, Netherlands
²Nevus 2000 France-Europe, Vandel, France;
³INSELM UMR_S910, Marseille, France
⁴Nevus Outreach, Bartlesville, Oklahoma, USA

Main purposes of the 2011 Expert Meeting in Tübingen

1. Establish permanent links between all existing patient groups (Figure 3) and spark the emergence of new and incipient associations
2. Introduce major international research and medical players to one another, and allow them to present their findings in a professional context
3. Allow patients and researchers to mingle and collaborate on the intention to design, populate and make use of an international registry

Patient group advances

In many Western countries, support groups are effective by:
- physically convening those affected and their families on a regular basis
- moderating online support groups about the disease (Yahoo!, Facebook, AceBoard, and others)
- staffing telephone hotlines
- providing accurate information through printed and online materials
- stimulating and actively raising funds for research

Representatives from each association determined the need in Tübingen to federate the above activities in blue.

The Global Naevus Initiative was created to develop a central clearinghouse website as a showcase for all worldwide associations, present and future (Figure 5), and for the prospective international large/giant CMN registry.

Research initiative advances

International registries for rare diseases are all the fashion. The Global Naevus Initiative has already mandated a 16-member international working group of doctors, scientists and a lawyer, to construct a prospective forerunner on the heels of experience with English and American patient group registries that no longer meet all research needs.

- In-person meeting of a quorum of the registry working group at the International Pigment Cell Conference in Bordeaux in September, 2011
- Pilot research grants awarded by three patient groups to scientists, some of whom have already secured additional funding from their first results
- Biobanking of blood and tissue samples in Europe and North America
- Peer reviewed publications by patient group-sponsored researchers²⁻¹

Conclusions

Since the 2011 Expert Meeting in 2011, patients, doctors and scientists are concretely working together around the world. The global registry and website are progressing toward completion, and should be announced in 2013 at the second meeting in a similar format.

Acknowledgments

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References


About Large Congenital Melanocytic Naevi (CMN)

A congenital melanocytic nevus (CMN; plural, naevi) with a projected adult diameter of over 10 or 20 cm is considered “large” or “giant”, respectively. While small (<1 cm) forms of this pigment-based birthmark are common, large forms occur at most in an estimated 20,000 births, and severe complications render syndromic forms of the disease even rarer (Figure 1).

Large and giant CMN are remarkable in addition to being rare; facial forms especially draw attention (Figure 2). Patients suffer from psychosocial difficulties, complex therapeutic management decisions, and the risk of neurological problems and malignant degeneration.

In the last two decades, support groups for LCMN were started from around the world.

Figure 1. A: Luca, from Italy, had an angiomatous presentation. B: (from Skin Cancer Recognition and Management 2nd ed. (2008) Robert A. Schwartz) This 17-year-old LCMN patient has already had two melanomas removed from her nevus; her dermatological management is typical of many patients.

Figure 2. Facial LCMN in children are similar around the world

Figure 3. Existing independent advocacy groups for patients with large/giant CMN as of May, 2011

Figure 4. Mockup of future GNI website

Figure 5. Patient groups that have been fostered since the creation of the Global Naevus Initiative

Figure 6. A: Tübingen meeting report published in a specialist journal². B: Example of registry entry information