

# INEQUITY ON NEUROFIBROMATOSIS TYPE 2 REGARDING KNOWLEDGE, AWARENESS AND LEVEL OF CARE: THE PARADIGMATIC ITALIAN SITUATION.

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Neurofibromatosis type 2 (NF2) is a spoiling and very neglected rare disease.

NF2 is multiple peripheral nerves, brain and spine tumors: schwannomas, meningiomas, ependymomas.

NF2 is caused by the genetic loss of the specific cellular tumor-suppressor-protein “merlin”. This means a predisposition for new and more tumors for life.

NF2 is a heartbreaking disease in teenagers and young adults: total deafness, facial palsy, loss of vision, speech problems (fig.1), multiple and severe neurological deficits.

NF2 patients often die in their twenties and thirties.



fig.1 NF2

**BUT NF2 is considered not as harmful as it is, because it is confused with a different and milder disease, Neurofibromatosis type 1.**

**The cause is the name, which is really a misnomer.**

“Neurofibromatosis” means “multiple neurofibromas as a hallmark”. On the contrary neurofibromas are only incidental in NF2.

“Type 2” means a relationship with Neurofibromatosis “type 1”.

That is not true! NF1 is a multi-systemic, with tens or hundreds of skin neurofibromas (fig. 2) and 10% tumor risk of different cancers from NF2 cancers.

NF2 is a 100% cancer and not a cutaneous disease.



fig.2 NF1

Such an intermingling was produced by very old confounding clinical reports, not accepted nowadays.

**Unfortunately the huge difference was not pointed out through renaming of NF2.**

Hence the false message---> **“NF1 and NF2: similar names, similar diseases”**

**NF2 is neglected even within specialized communities.**

NF2 is neglected in the European Rare Cancer Community.

Schwannomas are listed in the Rare Cancer List as sarcomas, soft tissues cancers. But NF2 patients die because of brain schwannomas!

NF2 is neglected by European Experts on Rare Cancers. They show interest in 3 ERNs, rare cancer networks, EURACAN, EuroBloodNet, PaedCan, (fig 3), while NF2 is in ERN GENTURIS.

**NF2 is neglected by many “NF” advocacy groups,**

theoretically dedicated equally to NF1 and NF2, but practically dedicated primarily to NF1.

**NF2 is neglected as timely accurate diagnosis and optimized care.**

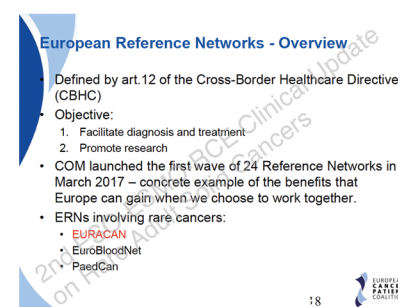
In the United Kingdom NF2 care has been nationally commissioned, with 4 NF2 clinics (Evans,2005).

English NF2 people don't suffer from high levels of anxiety or depression: they are clearly conscious to have the highest quality of care possible (Ferner, 2014).

Outside the UK, NF2 people are perfectly aware of how unlucky they are: insurance problems in USA, no NF2 clinics, very low level of awareness and competence in other countries.

Because of this inequality, our journey started in 2013 as the first NF2 advocacy group in Italy and in Europe:

Lega per la Neurofibromatosi 2/ League for Neurofibromatosis 2, a non profit organization.



## OUR CHALLENGES

In Italy the false message “NF1 and NF2: similar names, similar diseases” has brought to awful consequences, such as: A single identification code has been given for both NF1 and NF2, in Health National System (Law 279/2001). As consequence, a single official database as registry has been launched for “NF1-NF2” patients. Such a database can’t even count how many NF2 people are in Italy. **“If it can be measured, it can be managed”** is a main sentence in epidemiology!

NF clinics are usually directed by paediatricians, plastic surgeons and dermatologists, i.e. specialists intended for NF1 (while NF2 patients require neurologists, neurosurgeons and neuroradiologists in primis).

Visiting two of the most well-known NF2 centers in Europe (Manchester and Paris) has given us the chance to have their NF2 leaders support our needs. Indeed, a supporting letter from the top NF2 expert English geneticist Dr. Gareth Evans managed to stop the unstudied decision of the Italian Ministry of Health to shift **“NF1 - NF2 from tumoral to malformative diseases”** in 2015.

**NF1 is a malformative disease, NF2 is a cancer and not malformative disease!**

We are officially working as NF2 European Advocacy Group representative in ERN GENTURIS with the top expert NF2 clinics in Europe, on behalf of all European NF2 people (fig 4).

We are working through RareConnect.org to bring together NF2 patients who are left alone and isolated in their countries, treated by clinicians with no enough expertise in NF2.

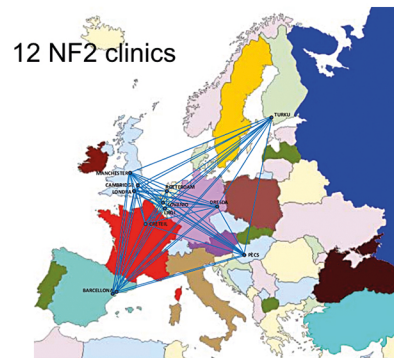


fig.4

## OUR PROPOSALS

A very close partnership with international lead clinicians is a first step, since they can be the best advocates and supporters for local patients' needs.

**“Stand on the shoulders of giants”.**

We believe in the power of a global NF2 alliance, with patients, top expert clinicians, researchers, with a single platform as point of reference, translated in different languages. That would be in our opinion the quickest and most authoritative way to raise awareness, knowledge and handle resistance.

Our dream is to have the dissemination of the UK NF2 model of care, and its NF2 registry, so powerful and precise to have changed the knowledge about NF2 until new diagnostic criteria (Baser,2002).

## REFERENCES

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