ERN: GENTURIS
Genetic Tumour Risk Syndromes

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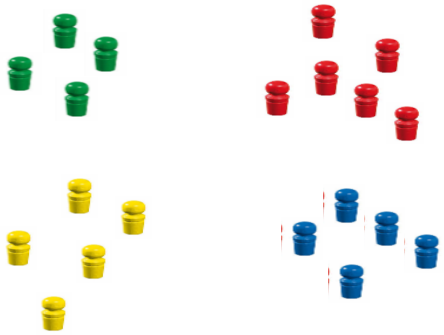
AIM of ERN GENTURIS

Improving the identification, diagnosis of a wide range of inherited syndromes predisposing for tumour development at any stage during life and prevention of cancer.

Healthcare	Patients	ERN	Healthcare providers
Patients at risk		Patient empowerment Creating general awareness	
Identification		Guidelines Teaching and training	All HCPs including GP's
Diagnosis	Multiple or one-stop investigation by GENTURIS expert team member	Guidelines Teaching Care, counseling Data- and biobanking Research	Clinical geneticists Molecular diagnostic geneticists In multidisciplinary team
Surveillance , Risk reducing surgery and treatment		Care Genetic counseling	Multidisciplinary HCP experts Depending on syndrome and manifestations
	Continued care within GENTURIS or referral to other ERN for rare manifestations or to general care for common manifestations	Guidelines Teaching Care, counseling Data- and biobanking Research Provide expertise and MDA to HCPs	

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Scope of diseases

Inherited syndromes predisposing for tumour development at any stage during life:

- Genetic cause is known
- Manifestations affect multiple organs
- Tumours can be benign or malignant
- Most malignant tumours are common cancers
- Identification and diagnosis is difficult
- Tumour prevention (surveillance and risk-reducing treatment) require a multidisciplinary team
- Personalized treatment based on germline mutations
- Healthy relatives at risk do not receive proper attention when index case is not recognized

Thematic groups

1. Neurofibromatosis

- NF1, NF2, Schwannomatosis

2. Lynch syndrome and polyposis

- Lynch Syndrome, Muir-Torre Syndrome
- (attenuated) familial adenomatous polyposis
- MYH-Associated Polyposis, DNA polymerase proofreading associated-polyposis
- NTLH1 associated polyposis, Turcot Syndrome
- Juvenile Polyposis Syndrome, Peutz-Jeghers syndrome
- Hereditary mixed polyposis, Serrated polyposis syndrome

3. Hereditary breast and ovarian cancer

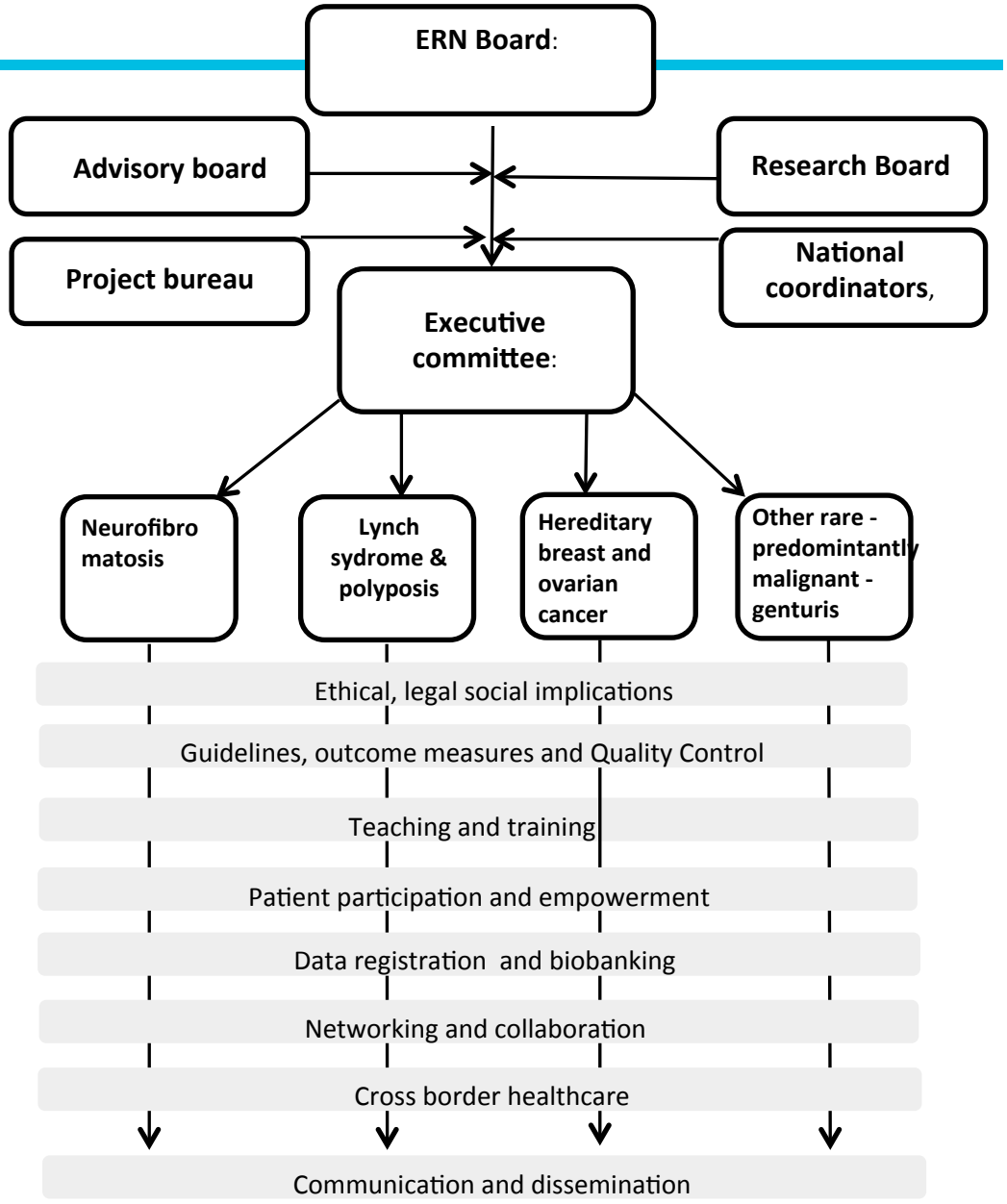
4. Other rare – predominantly malignant- genturis

- PTEN hamartoma tumour syndromes
- Li-Fraumeni Syndrome
- Birt-Hogg-Dubé Syndrome
- FAMMM
- Small Cell Carcinoma of the Ovary, Hypercalcaemic Type
- Hereditary diffuse gastric cancer (CDH1)

Disease expansion plan

- Paraganglioma
- Xeroderma Pigmentosa
- Carney Complex
- Hereditary Papillary Renal Carcinoma
- Ataxia-Telangiectasia
- Tuberous sclerosis
- Bloom syndrome
- Gastrointestinal polyposis syndromes
- Nevoid basal cell carcinoma syndrome
- Werner Syndrome
- Hereditary Leiomyomatosis and Renal Cell Cancer
- Newly detected genetic causes of cancer

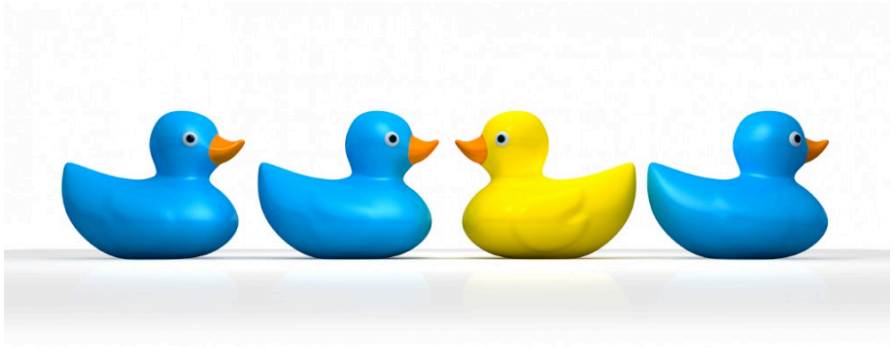
Structure of ERN GENTURIS



Why is this ERN necessary?

Unique Points:

- Predisposition of common cancers of various organ systems
- Prevention directed (including surveillance and risk-reducing treatment) and personalized treatment based on germline mutations.
- Relatives included



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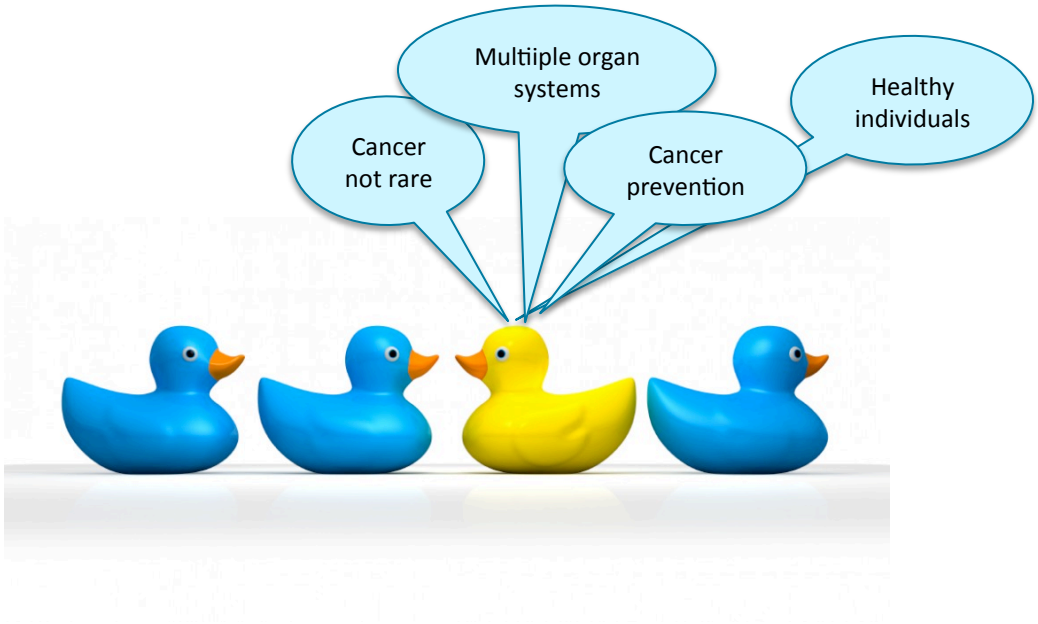


Multidisciplinary Professionals

- Inclusive, no competition, when meeting the criteria:
  - National endorsement
  - Minimum requirements
    - Numbers of patient in the thematic groups
    - Output: research, guidelines, boards.

- From at least 8 countries and 10 centers

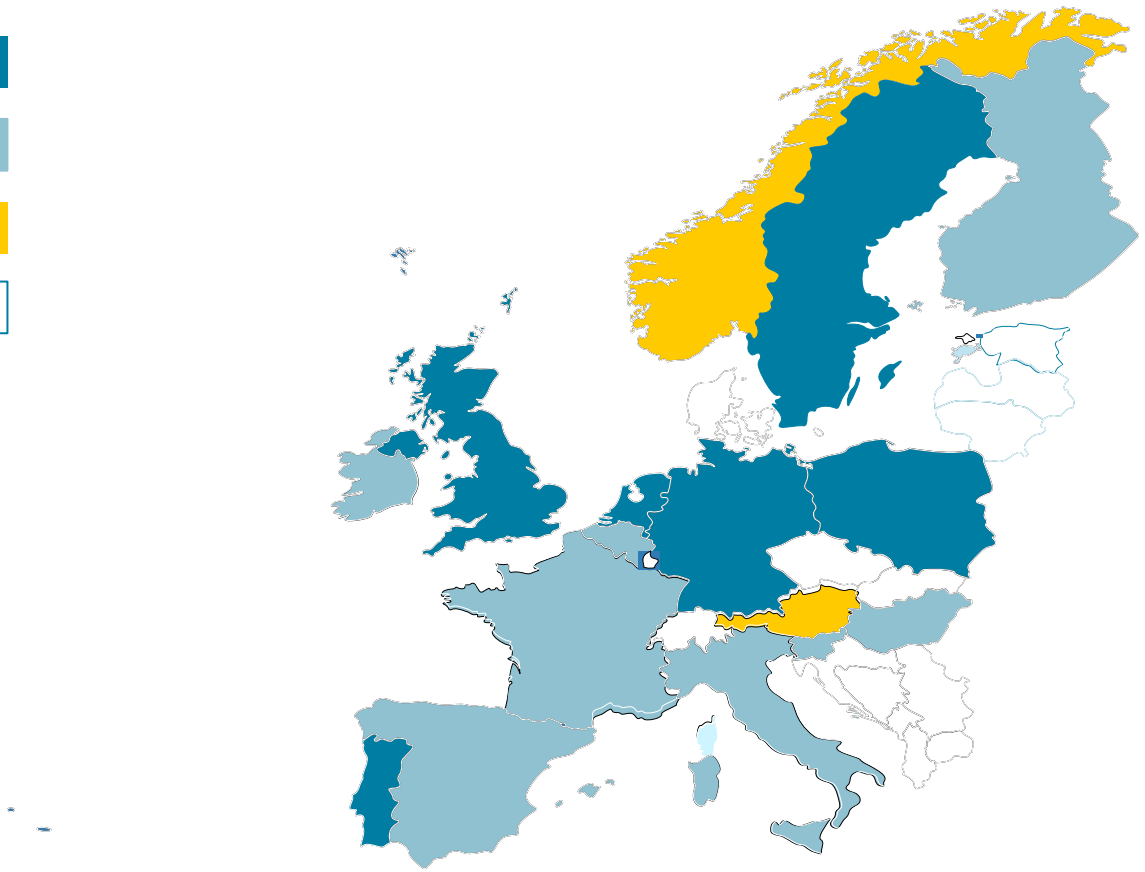
ERN: Genetic Tumour Risk Syndromes
Expertise centers (full members)

Endorsed HCP

Endorsement likely

Endorsement 2017?

No member yet



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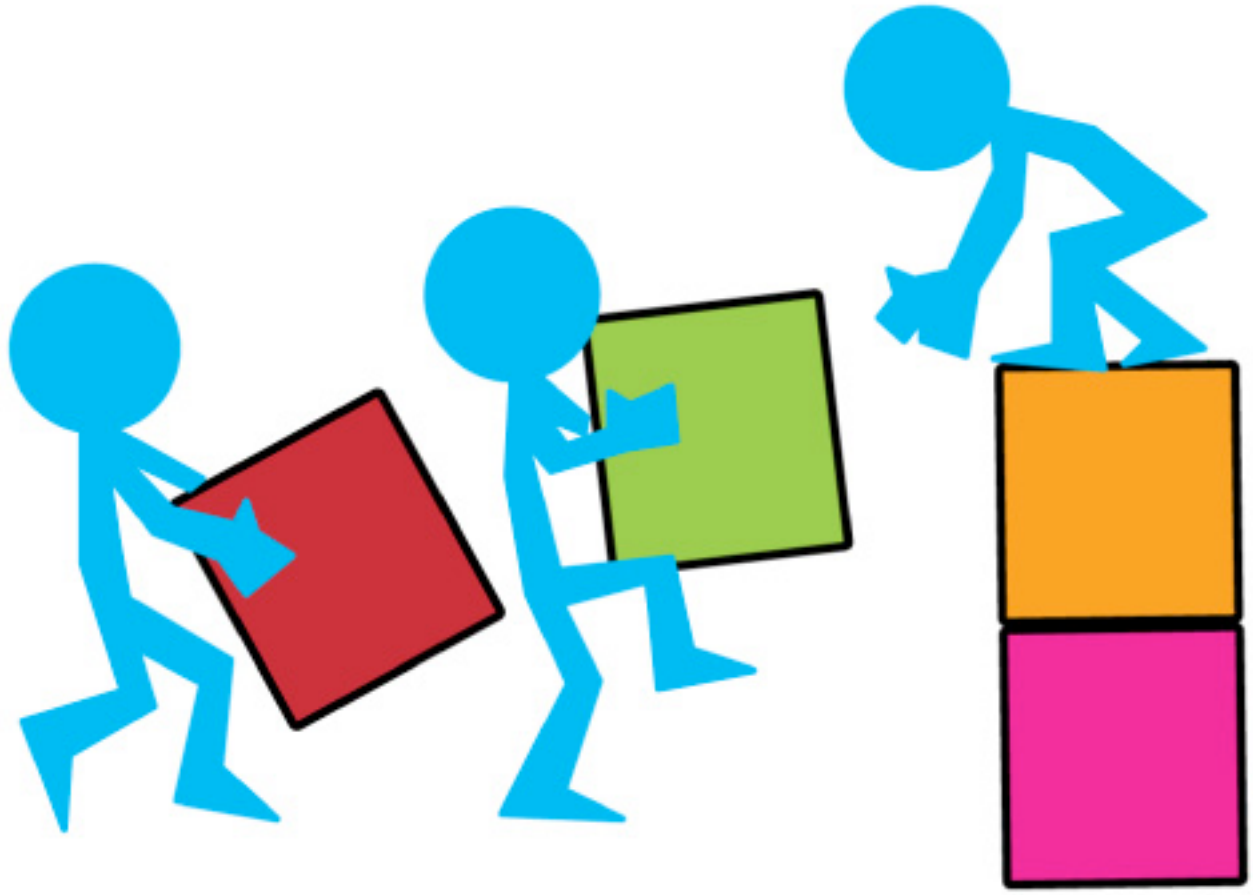

5. ERN Network application

- Introduction and inventory: (until 09.45u)
  What are the most important problems that need an answer now.

- Discussion in 4 groups (until 10.45u)
- Answers by per group in 15 minutes (until 12.00u)

  - What will be ready in 5 years from now?
  - Be practical, write in bullets (no complete sentences needed)

6. HCP application and self-assessment

- Introduction and inventory: (until 10.45u)
  What are the most important problems that need an answer now.

- Discussion in 4 groups (until 11.45u)
- Answers by per group in 15 minutes (until 13.00u)

  - What will be ready in 5 years from now?
  - Be practical, write in bullets (no complete sentences needed)