ERN: GENTURIS
Genetic Tumour Risk Syndromes

Prof Nicoline Hoogerbrugge MD, PhD
Coordinator
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

Identification

Diagnosis

Surveillance, Risk reducing surgery and treatment

Patients

Multiple or one-stop investigation by GENTURIS expert team member

Continued care within GENTURIS or referral to other ERN for rare manifestations or to general care for common manifestations

ERN

Patient empowerment
Creating general awareness

Guidelines
Teaching and training

Guidelines
Teaching
Care, counseling
Data- and biobanking
Research

Care
Genetic counseling

Guidelines
Teaching
Care, counseling
Data- and biobanking
Research

Provide expertise and MDA to HCPs

Healthcare providers

All HCPs including GP’s

Clinical geneticists
Molecular diagnostic geneticists
In multidisciplinary team

Multidisciplinary HCP experts
Depending on syndrome and manifestations

Radboudumc
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- genetris**
   - 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

- Executive committee:
  - Neurofibromatosis
  - Lynch syndrome & polyposis
  - Hereditary breast and ovarian cancer
  - Other rare - predominantly malignant - genturis

- Ethical, legal social implications
- Guidelines, outcome measures and Quality Control
- Teaching and training
- Patient participation and empowerment
- Data registration and biobanking
- Networking and collaboration
- Cross border healthcare
- Communication and dissemination

- ERN Board:
  - Advisory board
  - Research Board
  - National coordinators,
  - Project bureau

- Executives:
  - Advisory board
  - National coordinators,
  - Project bureau

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- Radboudumc
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
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)