European Reference Network: Genetic Tumour Risk Syndromes

ERN GENTURIS

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## Disclosure conflicts of interest

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<td>(potential) conflicts of interest</td>
<td>No</td>
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Presume you are 35 having polyps in your bowel

The surgeon tells you that you may have a genetic tumour risk syndrome including risk of multiple cancers (breast, bowel, thyroid and uterus) as well as autism (PHTS (Cowden syndrome) caused by a DNA defect in PTEN).

At that moment you need an expert!
Increased cancer risk PHTS (Cowden Syndrome)

Men

Women

Thyroid cancer

Bowel cancer

Breast cancer

Endometrial cancer

Melanoma?
Other features of PHTS (Cowden syndrome)
Presume you are 35 and at high risk of PHTS (Cowden syndrome)

- In a few European countries
  - you can be referred to a clinical geneticist or to an expert centre
  - there is a national guideline
  - you can find a patient advocacy group.
  - you can consent to a databank, biobank and research programs
Presume you are 35 and at high risk of a PTEN mutation

- In most EU countries
  - The surgeon has never heard of PHTS (Cowden syndrome) or PTEN mutations, and does not identify those at high risk.
  - There is a lack of cancer oriented clinical geneticists.
  - Little can be found written for patients in their own language
  - There are no guidelines.
Presume you are 35 and at high risk of a PTEN mutation

• In most EU countries
  • The surgeon has never heard of PHTS (Cowden syndrome) or PTEN mutations, and does not identify those at high risk.
  • There is a lack of cancer oriented clinical geneticists.
  • Little can be found written for patients in their own language
  • There are no guidelines.

• To share expert knowledge on complex or rare diseases, to speed up research, education and patient empowerment ERNs are being formed.
The ERN: European Reference Network

Networks of healthcare providers aiming at improving quality, and safety and access to highly specialised healthcare

Multidisciplinary approach (different specialities/areas of knowledge)

Need of cooperation:
- Scarcity knowledge
- Need education
- Complexity / high cost
- Effectiveness in the use of resources

"The knowledge travels, not the patient"
Share. Care. Cure.

Clinical virtual care

GENTURIS.eu
Thematic genturis groups

There are more than 40 different genturis

1. Neurofibromatosis

2. Lynch syndrome and polyposis

3. Hereditary breast and ovarian cancer

4. Other rare genturis e.g. PHTS
Opportunities ERN GENTURIS

Challenges when it comes to the identification, genetic testing, tumour prevention and treatment of patients with genturis:

1) Great majority of genturis patients are not yet identified
2) Large variation in clinical outcomes resulting in impaired prognosis and avoidable costs
3) Guidelines are lacking or implemented insufficiently
4) Almost no patient registries and biobanks
5) Limited research programs
6) Fragmented patient empowerment activities
ERN: networks of expert centers

- Diagnostics
- Treatment
- Prevention
- Research
- Teaching
ERN: Genetic Tumour Risk Syndromes
Expertise centers

Inclusive
Not Competitive!
Presume you are 35 and at high risk of PHTS (Cowden syndrome)

ERN GENTURIS

- Where ever you live in Europe you can find an expert for your rare disease.
- Medical experts all follow the latest EU guideline.
- Information can be found in your own language.
- You can participate in research to improve future care.
Conclusion

ERNs offer a unique opportunity that may have a significant impact on the health of many patients and families.