For additional informations and appointments with our outpatient clinic:

Dr. rer. medic. Heike Schackert-Görgens

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The Center for Familial Colorectal Cancer has an easy to use single contact number for all outpatient clinics including genetic counselling and surveillance and treatment of familial colorectal cancer:

For information:  +49 351 458 3598
9am to 4pm, Monday to Friday

for making appointments, cancel your appointment, update your contact details or obtain further information about your appointment.

The outpatient clinic for genetic counselling is open from 2pm to 5pm on Tuesdays.
Head: Prof. Dr. med. Andreas Tzschach.

The outpatient clinic for surveillance and treatment of familial colorectal cancer is open from 1pm to 3pm on Thursdays at the University Cancer Center (UCC, Building 32, 1st floor, Phone: +49 351 458 4500)
Head: Oberarzt Priv.-Doz. Dr. Steffen Pistorius.
Bethesda Guidelines for HNPCC

Suspicion of HNPCC arises if at least one of the following five criteria of the Bethesda guidelines is met (Umar, JNCI 2004). Consequently, tumor from the proband should be tested for mismatch repair deficiency:

1. Colorectal cancer diagnosed in a patient who is less than 50 years of age.
2. Presence of synchronous, metachronous colorectal, or other HNPCC-associated tumors*, regardless of age.
3. Colorectal cancer with the MSI-H† histology‡ diagnosed in a patient who is less than 60 years of age.§
4. Colorectal cancer diagnosed in one or more first-degree relatives with an HNPCC-related tumor, with one of the cancers being diagnosed under age 50 years.
5. Colorectal cancer diagnosed in two or more first- or second-degree relatives with HNPCC-related tumors, regardless of age.

If Bethesda guidelines are met, the proband gives written informed consent to molecular diagnostics after physician counselling. The proband is the affected individual through which a family with a genetic disorder is ascertained.

The optimal approach to molecular evaluation is immunohistochemical (IHC) analysis and/or microsatellite instability (MSI) testing of the proband’s tumor, to identify the knocked-out mismatch repair (MMR) gene. More than 50% of probands meeting Bethesda guidelines do not exhibit MMR deficiency in their tumor. Sequence analysis of one of the four mismatch repair genes is not recommended in these cases.

Immunohistochemical analysis of the tumor proceeds germline testing of one or two of the four MMR genes MLH1, MSH2, MSH6 or PMS2 depending on expression status in the tumor.

After mutation detection, the index person will be informed during genetic counselling. Relatives should then be referred for genetic counseling and predictive genetic testing.

A HNPCC specific surveillance is highly recommended for persons fulfilling one of the following criteria:

1. The proband meets Bethesda guidelines and the tumor is MMR deficient and no MMR gene mutation has been found.
2. The person is a relative of the proband (see 1.)
3. The person carries a pathogenic germline mutation in one of the four MMR genes.

Surveillance is recommended starting at the age of 25 or 5 years before the age of diagnosis in the youngest family member:

- Annual physical examination
- Annual abdominal sonography
- Annual total colonoscopy
- Annual gynecological examinations for endometrial and ovarian cancer (including transvaginal sonography)
- Annual gastroscopy
- Annual abdominal ultrasound
- Annual physical examination

* Hereditary nonpolyposis colorectal cancer (HNPCC) related tumors include colorectal, endometrial, stomach, ovarian, pancreas, uterine and renal pelvis, biliary tract, and brain (usually glioblastoma as seen in Turcot syndrome). Tumors, sebaceous gland adenomas and keratoacanthomas in Muir–Torre syndrome, and carcinomas of the small bowel.
† MSI-H microsatellite instability—high in tumors refers to changes in two or more of the five National Cancer Institute recommended panels of microsatellite markers.
‡ Presence of tumor infiltrating lymphocytes, Crohn's-like lymphocytic reaction, mucinous/signet-ring differentiation, or medullary growth pattern.
§ There was no consensus among the Workshop participants on whether to include the age criteria in guideline 3 above; participants voted to keep less than 60 years of age in the guidelines.

Diagnostic procedure in HNPCC-suspected cases

If Bethesda guidelines are met, the proband gives written informed consent to molecular diagnostics after physician counselling. The proband is the affected individual through which a family with a genetic disorder is ascertained.

The optimal approach to molecular evaluation is immunohistochemical (IHC) analysis and/or microsatellite instability (MSI) testing of the proband’s tumor, to identify the knocked-out mismatch repair (MMR) gene. More than 50% of probands meeting Bethesda guidelines do not exhibit MMR deficiency in their tumor. Sequence analysis of one of the four mismatch repair genes is not recommended in these cases.

Immunohistochemical analysis of the tumor proceeds germline testing of one or two of the four MMR genes MLH1, MSH2, MSH6 or PMS2 depending on expression status in the tumor.

After mutation detection, the index person will be informed during genetic counselling. Relatives should then be referred for genetic counseling and predictive genetic testing.

A HNPCC specific surveillance is highly recommended for persons fulfilling one of the following criteria:

1. The proband meets Bethesda guidelines and the tumor is MMR deficient and no MMR gene mutation has been found.
2. The person is a relative of the proband (see 1.)
3. The person carries a pathogenic germline mutation in one of the four MMR genes.

Surveillance is recommended starting at the age of 25 or 5 years before the age of diagnosis in the youngest family member:

- Annual physical examination
- Annual abdominal sonography
- Annual total colonoscopy
- Annual gynecological examinations for endometrial and ovarian cancer (including transvaginal sonography)
- Annual gastroscopy (starting at age 35)

If you have a suspicion of HNPCC …

. . . we offer the following state-of-the-art procedures:

1. You or your patient should make an appointment at our outpatient clinic. Please contact Dr. rer. medic. Heike Schackert-Görgens at +493514583873 or through the E-mail Heike.Goergens@uniklinikum-dresden.de. We will call you back soon thereafter.

2. In the case that the proband or a relative is not be able to visit our outpatient clinic, we will organize the diagnostic procedure with your help. In this case we shall need:

- Written informed consent of the proband.
- 10 ml of EDTA-Blood (no cooling required if sent to us within a few days)
- Clinical data: medical discharge report, histology report, colonoscopy and gastroscopy report, genetic counselling letter including the pedigree of the family
- Paraffin embedded tumor sample from the proband

Additionally, we offer counselling and treatment, including state-of-the-art genetic testing, for the following colorectal cancer syndromes:

- Adenomatous Polyposis Coli (APC) (Genes: APC, MUTYH)
- Peutz-Jeghers Syndrome (PJS) (Gene: STK11)
- Cowden Syndrome (Gene: PTEN)
- Familial Juvenile Polyposis (FJP) (Genes: MADH4, BMPR1A)