SCHELL GENETICS

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PRACTICE GUIDELINES

Dr Schell-Apacik is a consultant Clinical Geneticist who has been establishing his own private practice – Schell Genetics – based in London. He also started to work as an honorary consultant at St George's University Hospitals in London (NHS) in August 2021.

Dr Schell-Apacik has been working in genetics since 1990 specialising in Paediatric Genetics (childhood genetic disorders and syndromes) and has a sound knowledge in all aspects of clinical/ medical genetics. Furthermore, he carried out research on inherited brain malformations, and had the opportunity of working as a research fellow in the Children's Hospital in Philadelphia from 2000 to 2002. From 2005 to 2006 he had his own research project in the Children's Centre in Munich/Germany. Since 2007 he had his own Practice for Human Genetics in Berlin/Germany, before becoming the head of the Medicover Berlin-Westend Medical Care Unit.

Schell Genetics is an online practice, offering online genetic consultations primarily. The pivotal idea of the practice is that there has been a high demand in consultations with genetic specialists – even before the COVID pandemic.

This means that there is no "physical" practice where people come to, but an online way to connect and to get scheduled video sessions via a web platform. The patient material at this point is from the UK, but also from Germany and Europe, and could eventually at a later stage include patients also from other parts of the world. Even though Schell Genetics is a remote diagnostic service, there may be occasions where a face-to-face consultation with a patient (and their family if appropriate) may be considered more beneficial. In these cases, there is limited accessibility to a consulting room at No 10 Harley St in London.

Introduction

A genetic consultation and diagnostics are supposed to assist patients in making informed choices regarding the disease/ disorder in question, their health, or the health of their child, respectively. To what extent genetic consultations and diagnostics may be helpful depends on

the particular disease/ disorder and their personal specific questions. Both will define the topics in the consultation and the kind of genetic testing proposed. In the event of information being obtained during the consultation pointing to additional genetic issues the patient has not yet been aware of, their options will be discussed with them. It remains the patient's personal decision if and to which extent to proceed with a possible genetic diagnostic strategy.

Previous medical findings, clinical symptoms to date as well as the family history are a prerequisite for the genetic consultation and diagnostics.

A genetic consultation and diagnostics usually comprise:

- the clarification of the patient's personal reasons, motivation, and purpose for the consultation,
- a structured medical and family history including,
- an assessment and evaluation of all medical reports and results to date,
- an assessment of physical findings, and pictures taken for diagnostic purposes, if applicable
- initiating the genetic lab investigations/ testing,
- an assessment of specific genetic risks,
- a detailed Medical Genetics Report with the relevant findings including all lab results explaining possible health consequences and/ or treatment options, respectively.

Dr Schell-Apacik holds the opinion that patients are entitled to the available state-of-the art genetic tests and diagnostics and that access to them should be granted to optimise their care. Genetic diagnostics and testing will not be performed without an informed consent of the patient. It is their decision to make, and completely up to them whether to go ahead with a proposed genetic testing after the consultation. It is made clear that the relevance of proposed genetic strategies may depend on very personal factors. Equally, it remains the patient's decision to which extent to take the advice given regarding the results of the genetic diagnostics. Genetic testing may reveal findings with no known health effects to date. Dr Schell-Apacik would communicate such findings only if they were relevant for the enquiry or questions asked, respectively.

The patient is being made aware that it is impossible to obtain information on all conceivable genetic disorders or diseases influenced by genetic factors, respectively. They learn that it is equally not possible to assess or exclude all conceivable health risks for themselves, for family members and especially their children. In some cases, no risk estimates or likelihoods can be given for a particular disease/ disorder, striking features in the appearance of an individual (phenotypic abnormalities) or a developmental delay/ intellectual disability. Even if chromosome analyses and further genetic testing were uneventful, a genetic cause cannot be ruled out with certainty. In these instances, the genetic evaluation and assessment is based on scientific publications, literature cases and the comparison with other, likewise affected individuals. This also means that in the case of a pregnancy, there are cases – even after extensive genetic diagnostics and testing as well as thorough literature search – there are no definitive answers on whether the child will be healthy or not.

After completion of the genetic diagnostics patients and their referring physicians/ specialists/ GPs will be receiving a Medical Genetics Report with the most important topics of the consultation and the relevant findings including all lab results. The Medical Genetics Report also includes an assessment of the findings and recommendations enabling the referring colleague as well as other healthcare workers to specific follow-ups and tailored patient care.

Main Areas of Work

Dr Schell-Apacik has been offering online genetic consultations mainly for issues related to Paediatric Genetics (Childhood genetic disorders and syndromes), Oncogenetics (Cancer genetics), Reproductive Genetics and Neurogenetics.

- Paediatric Genetics
  A genetic cause may be considered in cases of
  - striking features in the appearance of a child (phenotypic abnormalities),
  - a developmental delay or intellectual disability,
  - an autism spectrum disorder,
  - a speech and language delay,
  - congenital anomalies or birth defects (e.g. of the extremities, inner organs),
  - a congenital hearing or visual impairment respectively,
  - a congenital metabolic disorder,
  - an inherited neurological or muscular disorder.

  Dr Schell-Apacik offers genetic consultation, diagnostics and testing in all cases of a suspected hereditary/ genetic disorder.

- Oncogenetics
  A genetic cause may be considered in the development of different cancers (e.g. breast cancer, ovarian cancer, colon cancer, pancreatic cancer, thyroid cancer, multiple endocrine neoplasia, retinoblastoma), or in the occurrence of multiple colon polyps.
  If a patient has been affected with a hereditary cancer syndrome not only this patient has an increased risk of developing another tumour (e.g. contralateral breast cancer, ovarian cancer, colon cancer, renal cancer, uterus cancer) but her/his relatives may also have an increased risk of developing tumours themselves.
  Dr Schell-Apacik offers genetic consultation, diagnostics and testing in all cases of a suspected hereditary cancer syndrome.

- Reproductive Genetics
  A genetic cause may be considered in cases of couples with problems in becoming pregnant or couples with habitual spontaneous abortions. These cases often undergo fertility treatment and in vitro fertilization (IVF, ICSI).
  A genetic cause may also be considered in cases of couples with pregnancy complication and fetal abnormalities.
  Dr Schell-Apacik offers genetic consultation, diagnostics and testing for couples with fertility issues and habitual spontaneous abortions as well as in cases with fetal abnormalities in prenatal diagnostics.

- Neurogenetics
  A genetic cause may be considered in the development of different neurological disorders (e.g. Hereditary Motor and Sensory Neuropathies (HMSN), Hereditary Spastic Paraplegia (HSP), Muscular Dystrophies (such as Duchenne, Becker), Hereditary Muscular Atrophies (such as SMA), Fragile X-Associated Tremor-/Ataxia Syndrome (FXTAS), Fragile X-Associated Neuropsychiatric Disorders (FXAND), Spinocerebellar Ataxias, Chorea Huntington, epilepsy syndromes)
  Dr Schell-Apacik offers genetic consultation, diagnostics and testing in all cases of a suspected hereditary neurological or muscular disorder as well as epilepsy syndromes.

But of course, Dr Schell-Apacik is happy to assist patients with all questions they might have on any other genetic issues.

Work Routine

In general, patients contact the practice online via email or the web portal.

They are being informed that genetic diagnostic testing is in general performed in relation to a clinical condition depending on the clinical symptoms, the severity, and the family history. They are told that to proceed with any request, a referral or formal request from a qualified specialist/

physician is usually required. In rare instances an online consultation can be performed without formal referral, but they would need to provide a GP/ clinician/ specialist in charge.

Patients are being asked to complete questionnaire and provide as much information as possible such as clinical reports, reports from specialists/ GP, medical history, standard photographs if applicable, family history including all individuals affected similarly indicating how they are related to them. To ensure confidentiality of all information provided, patients are invited to use the FTAPI platform[1] to submit their documents/ data securely.

After the evaluation of all submitted documents and the completed questionnaire, and depending on the given information, patients are being provided with a quotation for the online consultation services and payment details, the online genetic consultation services comprising an initial video session, a comprehensive Medical Genetics Report containing the most important topics of the consultation and the relevant findings including all lab results, and one follow-up session on a case to case bases if needed and required.

After accepting the quotation, patients are invited to an online appointment for genetic consultation, and a video session is being schedule via ZOOM. They are also asked to fill in a consent form for the consultation.

An initial session lasts usually approximately up to 1.5 to 2 hours (in exceptional cases, the genetic consultation session may take longer) depending on the complexity of the issues in question. Patients are being informed that the online consultations may be recorded for training and quality control purposes, and their right to object the recording if they wish to.

If patients are interested in genetic testing after the initial consultation session, they are being provided with a quotation for the recommended laboratory studies and asked to fill in an additional consent form for the genetic laboratory testing. Schell Genetics uses the services of CeGaT, a major genetic laboratory based in Germany.
Patients are being informed that they have the right to withdraw their consent at any stage without giving reasons, and that they also have the right, not to be informed about test results at all if they wish so (right not to know). In addition, patients are made aware of them having the right to halt the processing of the samples, and even demand them being discarded, at any time including all the results obtained heretofore.

Patients are being informed that the blood required can be drawn by their own local GP/ physician/ specialist or, alternatively, by local phlebotomy services. The blood draws should be performed ideally at the beginning of a week (Monday or Tuesday) to ensure the processing of the blood samples without any delay. They are being assisted by CeGaT directly with the shipping process and details[2].

After the laboratory studies are completed, Dr Schell-Apacik is provided by CeGaT with a detailed genetic analysis report.

The patient and their GP/ clinician/ specialist in charge are being provided with a comprehensive Medical Genetics Report as part of the genetic consultation services. The Medical Genetics Report summarises the most important topics of the consultation including the questions asked by the patient, their family history, their individual and medical history, a review of the available previous medical reports and results, and the relevant findings including all lab

[1] FTAPI is a German software provider located in Munich. FTAPI SecuTransfer is a software solution for end-to-end encrypted data exchange of large and sensitive data. https://www.ftapi.com/en/

[2] CeGaT sends patients a box via DHL Express, including a DHL Express return label, free of cost. This box contains everything the nurse/doctor needs to take the blood samples, including the shipping containers. Also, a special label is included with the package, identifying CeGaT as the responsible entity for the shipment and stating the contents as UN3373 Biological Substances CATEGORY B.

results. Part of the Medical Genetics report is also an assessment of all the genetic findings according to the current scientific knowledge, putting them in context of the symptoms and clinical presentation of the patient, and discussing the relevance of the results for the patient and possibly further family members. The evaluation and assessment of findings (lab results as well as phenotypic features of a patient) takes usually approximately 3 hours (in exceptional cases even longer) and is based on professional database information, scientific publications, literature cases and the comparison with other, likewise affected individuals.
This is followed by explaining possible health consequences and/ or treatment options, respectively, and recommendations such as specific and target-oriented follow-ups by their GP/ clinician/ specialist in charge as well as other healthcare workers if needed, refined treatment protocols and adjusted strategies depending on the results and the assessment. In this scope recurrence risks in the case of a pregnancy may be discussed, if appropriate, as well as the possibilities and limitations of prenatal genetic testing including options for prenatal diagnostics.

As part of the online genetic consultation services, patients are being offered a follow-up video session via ZOOM on a case to case bases if needed and required, to discuss with them the results in more details, explaining possible health consequences and their treatment options as well as recurrence risks and options for prenatal diagnostics, if appropriate. A follow-up session lasts usually approximately up to 1 to 1.5 hours depending on the findings, the outcome, the strategies proposed, and the questions patients might have. A follow-up session via ZOOM with the GP/ clinician/ specialist in charge might be considered on the patient's request or on the request of the colleague, the consent of the patient hereto provided.

Concluding Remarks

These PRACTICE GUIDELINES aim to give an overview of the concept and the work routine. It makes no claims to completeness. As with every work in progress, there might be a need to adept to unusual circumstances and unforeseen events.

Acknowledging that patients are entitled to the available state-of-the art genetic tests and diagnostics, patient care and the needs of the patient are paramount at every step of the genetic consultation services offered. It is designed to assist patients in making informed choices regarding the disease/ disorder in question and, ultimately, their health or the health of their child, respectively, and enabling the referring colleague as well as other healthcare workers to specific follow-ups and tailored patient care. However, it remains the patient's decision if and to which extent to take the advice and recommendations given.