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.

Dr. Schell-Apacik has been working in genetics since 1990, specialising in paediatric genetics (childhood genetic disorders and syndromes) and having 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 at the Children's Hospital in Philadelphia from 2000 to 2002. From 2005 to 2006, he had his own research project at 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 for consultations with genetic specialists, even before the COVID pandemic.

This means that there is no "physical" practice where people come, 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 occasionally also from Germany, Europe, and other parts of the world.

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 that information is 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 what extent to proceed with a possible genetic diagnostic strategy.

Previous medical findings, clinical symptoms to date, and the family history are prerequisites 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 the informed consent of the patient. It is their decision to make, and it is completely up to them whether to go ahead with the 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 what 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 to 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 for 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 distinctive features), 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 comparisons with other, similarly affected individuals. This also means that in the case of a pregnancy, there are cases where, even after extensive genetic diagnostics and testing as well as a 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 receive 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 provide 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
  - distinctive features in the appearance of a child (phenotypic distinctive features),
  - 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 or 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 suspected hereditary/ genetic disorders.

- 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 by a hereditary cancer syndrome, not only does this patient have 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 suspected hereditary cancer syndromes.

- Reproductive Genetics
  A genetic cause may be considered in cases of couples with problems becoming pregnant or couples with habitual spontaneous abortions. These cases often undergo fertility treatment and in vitro fertilisation (IVF, ICSI).
  A genetic cause may also be considered in cases of couples with pregnancy complications and foetal anomalies.
  Dr. Schell-Apacik offers genetic consultation, diagnostics and testing for couples with fertility issues and habitual spontaneous abortions, as well as for cases with foetal anomalies 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 suspected hereditary neurological or muscular disorders, as well as epilepsy syndromes.

But of course, Dr. Schell-Apacik is happy to assist patients with any questions they might have about 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 usually performed in relation to a clinical condition, depending on the clinical symptoms, the severity, and the family history. They are told that a referral or formal request from a qualified specialist or physician is preferable, even though it is not a prerequisite for a private consultation. In rare instances, a genetic consultation can be performed without a formal referral, but they would need to provide a GP/ clinician/ specialist in charge.

Patients are being asked to complete a questionnaire and provide as much information as possible, such as clinical reports, reports from specialists/ GP, medical history, standard photographs if applicable, and family history, including all similarly affected individuals, indicating how they are related to them. To ensure the confidentiality of all information provided, patients are invited to use a dedicated platform 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, and they are invited to book an online appointment for genetic consultation. The online genetic consultation services comprise an initial video session via Zoom, 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-by-case basis if needed and required. Patients are also asked to fill out a consent form for the consultation.

An initial session usually lasts 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 that they have the right to object to 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 out an additional consent form for the genetic laboratory testing.

Schell Genetics uses the services of two genetic laboratories: CeGaT, based in Germany (https://cegat.com/), and Blueprint Genetics, based in Finland (https://blueprintgenetics.com/), both laboratories offering a wide range of state-of-the-art genetic tests, whereby CeGaT favours blood samples and Blueprint Genetics buccal swab samples.
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 their right to halt the processing of the samples and even demand that they be discarded at any time, including all the results obtained heretofore.
If blood samples are required for testing, patients are being informed that the blood 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 the week (Monday or Tuesday) to ensure the processing of the blood samples without any delay.
If buccal swab samples are required for testing, patients are being informed that the laboratory will send them a buccal swab sample collection kit via mail, and they are being asked to follow the instructions provided by the lab carefully.
Patients are being assisted directly by the respective genetic laboratory with the shipping details and process[1].

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

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 results. Part of the Medical Genetics report is also an assessment of all the genetic findings according to 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) usually takes approximately three hours (in exceptional cases even longer) and is based on professional database information, scientific publications, literature cases, and the comparison with other, similarly 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-by-case basis if needed and required, to discuss with them the results in more detail, explaining possible health consequences and their treatment options, as well as recurrence risks and options for prenatal diagnostics, if appropriate. A follow-up session usually lasts approximately 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

[1] Both CeGaT and Blueprint Genetics send patients a box via DHL Express. This box contains everything the patients need for the buccal swab samples, or the nurse/doctor needs to take the blood samples, including the shipping containers.

with the GP/ clinician/ specialist in charge might be considered on the patient's request or on the request of the colleague, with 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 adapt to unusual circumstances and unforeseen events.

Recognising 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 to enable the referring colleague as well as other healthcare workers to provide specific follow-ups and tailored patient care. However, it remains the patient's decision whether and to what extent to take the advice and recommendations given.

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