CLINICAL GENETICS SERVICES, CENTRE FOR HUMAN GENETICS

1. What is Clinical Genetics?

Clinical Genetics is a medical specialty that deals with conditions caused by changes in genes or chromosomes. It helps in diagnosing inherited disorders, understanding health risks, and guiding treatment, prevention, and family planning.

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2. Who should visit the Clinical Genetics clinic?

You may benefit from a **genetics consultation if you or your family member has:**

- Unusual facial features or birth anomalies
- Ambiguous genitalia
- Abnormal newborn screening
- Abnormal head shape, cleft lip/palate
- Suspected metabolic disorder from infancy
- Inherited hearing loss, blindness, heart problems
- Inherited blood disorders eg; thalassemia, sickle cell anemia
- Unusual growth patterns (short stature or tall stature)
- Developmental delay or Intellectual disability or autism (testing in particular)
- A known genetic disorder in the family
- Early-onset cancers or multiple cancers in the family
- Couple/ parents married in relation (consanguineous marriage)
- Repeated pregnancy losses or infertility
- Prior to planning Assisted Reproductive Technique/ In vitro Fertilization (ART/IVF)

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3. Do I need a referral to visit the Genetics clinic?

Not necessarily. You can contact us directly if concerned about possibility of a genetic condition in the family. Sometimes you may be referred by your treating doctor, relative or even a well-wisher.

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4. How can I book an appointment?

Appointments can be booked through phone call or email.

Mobile number: **09448894219** Email: geneticappointments@gmail.com

We provide direct consultation as well as online consultation, the details of which will be shared with you during the initial conversation with appointment team.

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5. How long will it take for an appointment?

If it is an ongoing pregnancy, it will be prioritised. We try our best to see you in 2-3 weeks' time. You will be updated about the tentative appointment date from our appointment team.

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6. How should I prepare for my appointment?

If possible, bring:

- All available previous medical records and test reports, X-rays, CT/MRI films
- Pregnancy and birth history
- Information about affected family members (who, what condition, age of diagnosis)- photographs, medical documents

You may be asked to share all these documents over e-mail id if it is an online consultation.

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7. What happens during a genetics consultation?

During the visit:

- The project assistant/genetic counsellor will take a detailed medical and family history which will be reviewed by the doctor
- Previous medical records will be reviewed
- A detailed physical examination will be done
- Genetic tests will be advised if needed
- Counselling will be provided about diagnosis, treatment, and future planning

The consultation is usually longer than a routine doctor visit.

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8. What is genetic counselling?

Genetic counselling helps individuals and families to:

- Understand genetic conditions

- Know the chances of a condition occurring or recurring
- Make informed medical and reproductive decisions
- Cope with emotional and psychological aspects

It does **not** force any decision—it only provides information and support based on the clinical details so that the family can take appropriate decision about next step.

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9. What are genetic tests?

Genetic tests are laboratory tests that examine genes or chromosomes to identify changes that may cause disease. These are commonly done in blood samples but can be done in other samples also.

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10. Are genetic tests painful or risky?

Most genetic tests require only a blood sample and is generally safe. In special situations (such as during pregnancy), other procedures like invasive prenatal testing (Chorionic Villous Sampling -CVS/ Amniocentesis) may be discussed, along with their risks and benefits.

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11. Is genetic testing necessary for everyone?

No. Genetic testing is advised only when it is medically indicated. The doctor will explain:

- Why a test is needed
- What information it can provide and its application in relation to monitoring, treatment, planning future pregnancies
- Its limitations

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12. Can genetic disorders be treated or cured?

Some genetic conditions can be treated or managed with:

- Medications
- Dietary changes
- Hormone therapy
- Surgery
- Supportive therapies

While not all genetic conditions can be cured, early diagnosis can greatly improve quality of life and outcomes.

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13. Are genetic conditions always inherited?

No. Some genetic conditions are inherited from parents, while others occur for the first time in an individual due to new genetic changes.

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14. If one child has a genetic disorder, will future children also be affected?

The risk depends on the underlying gene and its pattern of inheritance. These are explained to the family during a genetics consultation which can help estimate the chances and discuss options for future pregnancies.

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15. Can genetic testing be done during pregnancy?

Yes. Prenatal genetic testing can be done in certain situations to assess the health of the fetus. These tests are offered only after proper genetic counselling, evaluation and with consent.

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16. Will my genetic information be kept confidential?

Yes, absolutely. Genetic information is treated as strictly confidential and the test are done you're your consent. The test results are shared only to the couple/ affected individual/ parents in case of a minor child, according to hospital policies and ethical guidelines.

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17. How long does it take to get genetic test results?

The time varies depending on the test:

- Some results are available in a weeks' time and the more detailed chromosomal and gene related studies take about 4-5 weeks' time.

You will be informed about the approximate timeline during the counselling session.

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18. Are genetic tests expensive?

The cost depends on the type of test required. The cost of these genetic tests has come down over the last few years. The approximate cost will be informed to you during the counselling session.

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19. Can children be seen in the Genetics clinic?

Yes. Clinical Genetics services are available for couples who are planning pregnancy or having an ongoing pregnancy, newborn babies, children and adults.

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20. Do genetic conditions mean disability?

Not always. Genetic conditions vary widely. Some individuals live completely normal lives, while others may need medical or developmental support for a better quality of life.

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21. Will one visit be enough?

After the initial visit, a follow-up appointment is generally required to discuss about the genetic test results once these are available. Further follow-up visits depend on the underlying genetic condition. Some of the genetic conditions require annual follow-up and some require more frequent visits and investigations. This will be planned during the initial follow-up session.

You may be enrolled to the multispecialty clinic/annual follow-up clinic once the diagnosis is confirmed. eg. Gaucher disease, Mucopolysaccharidosis (MPS), Neurofibromatosis (NF1) etc.

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22. What should I do if I have any more doubts?

Kindly contact us at **09448894219** and you will be assisted.
