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 CUHK Medical Centre
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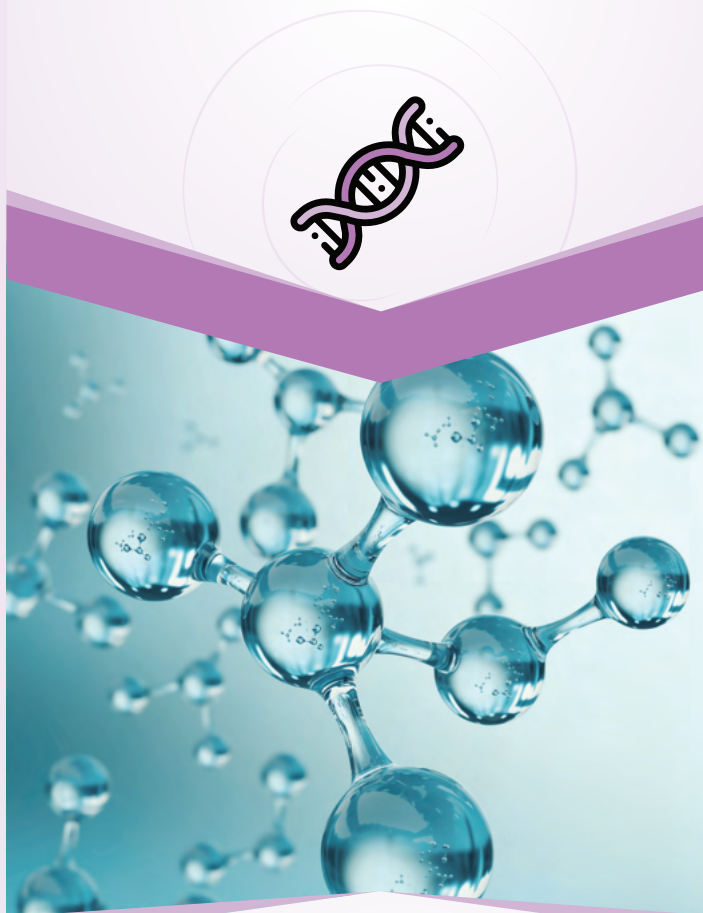
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CUHKMC/H/L/GG/INTRO/2022/4

香港中文大學醫院 醫學遺傳科門診

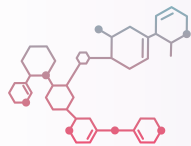
CUHK Medical Centre Clinical Genetics Clinic



香港中文大學醫院
CUHK
Medical Centre

本院的臨床遺傳科醫生及跨專業團隊，可為多種疾病如單基因疾病、發展遲緩、腦癱症、先天性異常、自閉症、癌症、退化性疾病和代謝功能障礙等的病人，進行基因診斷和提供具有質素的遺傳諮詢服務。

Our clinical geneticist and multidisciplinary team offer comprehensive services to advance genetic diagnosis and quality counseling service for our patients with Mendelian disorders, developmental delay, epilepsy, congenital malformations, autism, cancers, degenerative disorders, and inborn error of metabolism disorders.



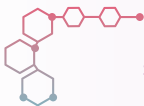
服務 Services

我們提供的醫療程序

遺傳諮詢服務會提供一站式綜合臨床遺傳評估，對遺傳病和罕見疾病進行基因測試和診斷。醫護人員會進行遺傳諮詢，以幫助患有遺傳性疾病或罕見疾病的患者得到基因確診，幫助患者家庭了解遺傳性質、遺傳方式、家庭中復發的風險以及預防方法；以便在計劃生育方面做出知情同意的決定。

Procedures that we offer

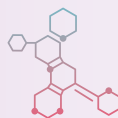
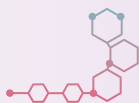
The clinical genetics clinic offers one-stop comprehensive clinical genetics service including genetics assessment, diagnostics genetics and genomic testing on genetic diseases, and pre-/post- genetics test counselling. Genetic counselling will help patients or families with a genetic disease or a rare disease to understand the necessity of the genetic test, the limitation of test, the nature of the disease, hereditary nature, the mode of inheritance, the risk of occurrence of the disease within the family, and the means of prevention; so that informed decisions are made in family planning.



基因測試 Genetic Testing

基因測試會從患者採集少量血液或組織樣本，測試樣品細胞中包含的DNA（基因），以檢測當中任何有致病風險的變異或突變。視乎進行那種基因測試，基因測試報告需時數天至數週。

Genetic testing usually involves taking a small sample of blood or tissue from the client. DNA (genes) contained in the cells from the sample can be tested to detect variation or mutation that are at risk of developing any genetic condition. Depending on what kind of genetic test is done, the availability of genetic result takes from days to weeks.



服務對象 Target Group

兒科疾病 Paediatric Conditions

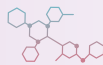
- 先天缺陷
- 經新生兒篩查發現疑似患有遺傳病
- 發展遲緩
- 自閉症譜系障礙
- 專注力不足/過度活躍症
- 智力障礙
- 腦癱症
- 其他系統功能障礙（聽力損失、皮膚、心臟、腎臟等）
- Birth defect
- Suspected genetic disease by newborn screening
- Developmental delay
- Autistic spectrum disorder
- Attention deficit hyperactivity disorder
- Intellectual disability
- Epilepsy
- Other system dysfunction (hearing loss, skin, heart, kidney disorder, and etc.)

生殖健康情況 Reproductive Health Issues

- 想進行基因攜帶篩查的女士
- 經歷過多次流產或嬰兒夭折的婦女
- 表親或近親的夫婦
- 超聲波檢查或血液檢測結果高風險出現併發症或先天缺陷的孕婦
- Women who are interested in genetic carrier screening
- Women who have experienced multiple pregnancy losses or babies who died in infancy
- Couples who are first cousins or other close blood relatives
- Pregnant women whose ultrasound examinations or blood testing indicate that their pregnancy may be at increased risk for certain complications or birth defects

家族病史 Family History

- 患有智力發展障礙、發展遲緩、遺傳性疾病或先天性缺陷
- 因已知或未知的醫療狀況而早年離世
- 患有成年早期發病的心血管疾病、認知障礙症或癌症等健康問題
- 希望檢測或了解更多關於在其族裔群體中高頻率的遺傳病的夫婦
- Intellectual disability, developmental delay, an inherited disorder, or a birth defect
- Early deaths due to known or unknown medical conditions
- Adult-onset health conditions such as cardiovascular disease, dementia, or cancer, particularly if onset is early in adulthood
- Couples who would like to test or obtain more information about genetic conditions that occur with higher frequency in their ethnic group



測試 Test

核型 Karyotyping

胎兒測序 Fetalseq V1.0

擴展性攜帶者篩查 Expanded Carrier Screening

基因組測序（指定基因組） Gene Panel (Various panels)

全外顯子體測序 Whole Exome Sequencing (WES)

脆性X攜帶者篩查 Fragile X carrier screening