CTNNB1 綜合症 (CTNNB1 Syndrome)

CTNNB1 SYNDROME

CTNNB1 syndrome is a newly recognised condition, first reported in 2012 for its association with developmental delay and intellectual disability. It occurs when there is mutation in one of the two copies of CTNNB1 gene (located on chromosome 3) causing loss of function of the gene.

MANIFESTATIONS - wide spectrum among patients

Developmental delay

Intellectual disability, learning difficulties

Low muscle tone in trunk but increased muscle tone over lower limbs, scoliosis

Microcephaly

Speech impairment

Vision problems: squint, hyperopia, myopia

Special facial features: broad nasal tip, long philtrum and thin upper lip

Sleep problem, autistic features, ADHD, aggression, behavioural problems

Startle response: very sensitive to sound

A minority have abnormal MRI of brain or spine

CTNNB1 綜合症是什麼?

- CTNNB1 綜合症是近年發現的病症，於 2012 年才首次有這病症的報告
- CTNNB1 基因位於第三對染色體，當兩個 CTNNB1 基因其中一個出現突變而失去功能，便會出現 CTNNB1 綜合症
- 在香港只得極少確診個案，估計全世界暫時也只有數百個確診個案

博軒可能是香港首名確診患 CTNNB1 綜合症的病人，有關他的故事請看以下的微電影【不一樣的哥哥】：

https://www.youtube.com/watch?v=3LRp0Njfzsl

症狀

症狀的種類和嚴重程度在患者間有很大差異

- 發展遲緩，智能障礙，語言障礙，學習困難
- 肌肉張力異常：軀幹肌肉張力低但雙腳張力高，引致行動困難、脊柱側彎、雙腳變形、腳跟韌帶過緊等情況
- 小頭症
- 視力問題：如遠視、近視和斜視
- 特殊的面貌特徵，例如鼻翼較小、人中長而淺、上唇較薄等等
- 部分患者有行為問題、睡眠問題、對聲音敏感引致驚愕反應、自閉症、專注力不足及過度活躍等症狀
- 大部分患者的腦部磁力共振正常，但小部分人的胼胝體或腦幹發展不全，腦回異常，腦室漲大，髓鞘形成異常等等，亦有些患者的脊髓磁力共振異常

尋找 CTNNB1 綜合症的同伴

CTNNB1 綜合症 (CTNNB1 Syndrome)

DIAGNOSIS

By detection of the mutated gene through Gene Sequencing. With increasing availability of gene sequencing, it is expected that more patients will be diagnosed in the future.

CHANCE OF HAVING AFFECTED SIBLINGS

As CTNNB1 gene mutation is mostly de novo, the risk of having a sibling with CTNNB1 syndrome is very low. Yet there is a rare case of CTNNB1 syndrome affecting two children in a family.

MANAGEMENT

At the moment, there is no cure for CTNNB1 syndrome.

Most patients are followed up by Geneticists, Paediatricians, Neurologists or Ophthalmologists

Combination of special educational support, speech therapy, occupational therapy, physiotherapy, hydrotherapy and hippotherapy may be helpful.

Some need medications for muscle tone, sleep and psychiatric symptoms. Some need surgery for squint correction or improving muscle tone.

Possibility of gene therapy is yet to be explored.

診斷

CTNNB1 綜合症需要經基因排序才能診斷，相信隨著基因排序日漸普及，日後會發現更多確診個案

遺傳風險

CTNNB1 綜合症的基因突變絕大部分為新生突變 (de novo mutation) ，並非由父母遺傳，所以患者的兄弟姊妹患病的風險與一般人無異，但亦有姊妹一同患病的罕見個案

治療

- 目前仍然未有根治 CTNNB1 綜合症的方法
- 及早診斷可以幫助患者監察以上症狀和進行適切的治療
- 一般由遺傳科、兒科、腦科或眼科醫生跟進
- 特殊教育支援、物理治療、職業治療、言語治療、水療、馬術治療也可對病者有幫助
- 減低肌肉張力的藥物、改善睡眠及精神症狀的藥物
- 部分病者需接受眼科和改善肌肉張力的手術
- 未來能否運用基因治療仍有待探索

CTNNB1 Syndrome 的資訊和聯絡:

- CTNNB1 Syndrome Awareness Worldwide (CSAW)
  https:// www.ctnnb1.org

CSAW 是 CTNNB1 綜合症的家長組織，旨在為新確診的家庭及醫療護人員提供資訊

- Unique CTNNB1 Syndrome disorder guide
- Facebook 有 CTNNB1 綜合症的家長群組，現有超過一百個來自世界各地的家庭，包括由幾個月大至 36 歲的病友，暫時群組內只有一個香港家庭
- 由於 CTNNB1 綜合症在香港尚未有病人組織，如有需要可透過香港黏多醣症暨罕有遺傳病互助小組聯絡

電郵: hkmpsinfo@gmail.com

電話: (852) 2794-3010

網址: mps.org.hk

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