HIE-CP Genomics

Tajul Arifin Tajudin

KPJ Puteri Specialist Hospital/Hospital Sultan Ismail, Johor Bahru, Johor

SUMMARY

Neonatal hypoxic-ischemic encephalopathy (HIE) occurs in approximately 1.5 per 1,000 live births in high-income countries and is even more prevalent in middle- and low-income regions. One of the most common complications resulting from hypoxic-ischemic encephalopathy is cerebral palsy. Cerebral palsy is a neuromotor disorder that primarily affects movement, muscle tone, and posture. It results from brain injuries sustained during the prenatal or neonatal periods. While the primary brain injury remains unchanged, children with cerebral palsy may experience a range of secondary conditions over time, potentially affecting their functional abilities and overall quality of life.

There are several early indicators that can help identify and suspect cerebral palsy in infants and young children. Developmental delays, such as difficulties with rolling over, sitting unsupported, or crawling, are common. Abnormal muscle tone, which can manifest as either excessive stiffness (hypertonia) or unusual limpness (hypotonia), is another key sign. Children may also display unusual postures, including asymmetry in movement or positioning. Additionally, premature hand preference – favouring one hand noticeably before 12 months of age – can signal possible hemiplegia.

Although the above can be indicative of cerebral palsy, they are not exclusive to cases linked to hypoxic-ischemic encephalopathy (HIE). Various other conditions, including genetic disorders, metabolic diseases, or structural brain abnormalities, can present with similar characteristics. A thorough medical evaluation, including neuroimaging and clinical assessments, is essential for determining the underlying cause and ensuring appropriate intervention. It is of utmost importance to know when neonatal encephalopathy is not secondary to HIE.

Cerebral palsy (CP) is a complex neurodevelopmental disorder with multifactorial origins, including genetic contributions that are increasingly being recognised. Advances in genomic research have provided new insights into the potential genetic predisposition to CP, uncovering mutations and variations associated with motor function and neurodevelopment.

In my presentation, I will not only discuss the well-established aspects of the hypoxic-ischemic encephalopathy (HIE) and cerebral palsy relationship but also shed light on overlooked early indicators in movement and posture among children at risk of developing cerebral palsy. To conclude, I will also explore the intersection of cerebral palsy and genomics, highlighting recent discoveries that enhance our understanding of the disorder's genetic aetiology and emerging pathways for personalised therapeutic interventions.