ECRONICON OPEN ACCESS

EC PAEDIATRICS
Opinion

Stroke-Like Episodes Secondary to Metabolic Disease

Jorge Sales Marques*

Pediatric Department, Centro Hospitalar Conde S. Januário, Macau, China

***Corresponding Author:** Jorge Sales Marques, Pediatric Department, Centro Hospitalar Conde S. Januário, Macau, China.

Received: November 08, 2018; **Published:** December 26, 2018

Stroke-like episodes are rare in childhood and adolescence.

Sometimes, without any special reason, we see children and adolescent in intensive care unit with stroke-like episodes.

There are several causes responsible for these episodes, such as cerebral malformations, infections, injuries, heart diseases, coagulopathies, drug use and prematurity.

Within the metabolic causes, some situations that may present this picture are: MELAS (mitochondrial encephalomyopathy lactic acidosis, stroke-like episodes), classical homocystinuria and Fabry disease.

How we differentiate them?

MELAS

The onset of the symptoms are between 5 - 15 years of age.

The main presentation are: headache, vomiting, convulsions, diabetes, cardiomyopathy, short stature, exercise intolerance, stroke-like, progressive external ophthalmoplegia (PEO) and sensorineural hearing loss.

The muscle biopsy showed ragged red fibres (RRF). Around 80% of mt DNA mutations are m.3243A>G(MT-TLI), mostly maternal inheritance.

Classical Homocystinuria

Is an autosomal recessive disorder with cystathionine beta synthase deficiency.

The patients are asymptomatic until 3 years old. Later, they showed inferior deviation of the crystalline, cataracts, retina dislocation, marfanoid phenotype, psychomotor delay, scoliosis and kyphosis, aorta degeneration. The stroke-like episodes appear at 15 yrs of age and is caused by thromboembolism.

Blood tests, revealed plasma amino acid with high level of methionine and homocysteine and reduced cysteine. The nitroprusside test is positive. The molecular study confirm the diagnosis with the determination of CBS gene.

Fabry disease

Fabry disease is an inherited X-linked autosomal recessive disease caused by an inability to produce an enzyme called alpha-galactosidase or alpha-GAL

Without this enzyme, a kind of fat called globotriaosylceramide or GL-3, which should be removed from the body, remains in the cells.

The result is the accumulation of this material in the blood vessels, leading to malfunctioning of the kidneys, heart and brain.

The symptoms of Fabry's disease appear between 6 and 9 years of age.

Pain is considered the first and most common of all symptoms. Most people with the disease have two types of pain: acroparesthesia and "Fabry crises". The acroparesthesia - pain that basically affects the hands and feet. It is described as a burning pain accompanied by a tingling that can be intermittent or daily. "Fabry crises" - episodes of intense, unbearable and burning pain, initially in hands and feet and radiating to other parts of the body. They can last from a few minutes to a few days.

Others symptoms included: 1- Hypohidrosis/anhydrosis: frequent fevers, overheating with physical exercise and intolerance to hot weather. 2- Angiokeratoma: characteristic purple-red rash - is the most visible sign of Fabry's disease - are found from the navel to the knees and, in some cases, only on the elbows or knees. 3- Cornea verticillate: similar to the rays of a bicycle wheel (does not affect the vision) - ophthalmoscopy of slit lamp. 4- Epigastric pain, diarrhea and nausea after meal, renal insufficiency due to excess proteinuria, cardiomyopathy, defective heart valves, arrhythmias, heart failure, dizziness, vertigo, headache, depression and stroke-like episodes.

The confirmation of the diagnosis is with the determination of GLA gene.

The important of the diagnosis is the possibility to do genetic counseling and prenatal diagnosis for the couple in their future pregnancy.

Fabry disease can be treated by enzyme replace therapy when is detected.

Stroke-like episodes are situations that really need a complete approach by the physician. If we can find what was the really cause of the episode, we can reduce the risk of sequel and avoid new recurrence in the future [1-3].

Bibliography

1. Jorge Sales Marques. "Screening for Pompe, Fabry, Gaucher and Mucopolysaccharidosis Type 1 (MPS 1): A Clinical and Laboratory Overview". *EC Paediatrics* 2.6 (2016): 268-271.

2. Jorge Sales Marques. "Stroke in Newborns and Infants: What Etiology?" *EC Paediatrics* 6.3 (2017): 53.

3. Jorge Sales Marques., *et al*. "Dried Blood Spot Samples for Pompe, Fabry, Gaucher and Mucopolysacharidosis (Mps): Our First Year Experience". *Archives of Pediatrics and Neonatology* 1.1 (2018): 13-16.

Volume 8 Issue 1 January 2019