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Image and Learning

Esophageal Dysmotility in Gillespie Syndrome

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A 16-year-old girl presented with dysphagia and heartburn for 10 years. She was diagnosed with Gillespie syndrome at the age of 1 year. Neurologic findings were represented by bilateral aniridia, strabismus, ataxia and cognitive impairment. Karyotype was normal (46, XX).

The upper digestive endoscopy disclosed an esophageal dilation and a 5 cm sized Barrett's esophagus confirmed by biopsy. High-resolution manometry showed aperistalsis and a non-detectable lower esophageal sphincter due to severe hypotonia (Figure), corresponding to absent peristalsis on the Chicago classification.[1] Ambulatory 24 hours pH monitoring disclosed a pathological acid reflux (total % time pH < 4: 36%, DeMeester score = 149).

Gillespie syndrome is a very rare disease described firstly in 1965. It is defined by the triad of cerebellar ataxia, aniridia and mental deficiency.[2] Associated manifestations have been infrequently described.[3,4] However, esophageal involvement has never been reported.

Although the presented association between Gillespie syndrome and esophageal dysmotility may be incidental, there is also a possibility that esophageal dysmotility could be a true sign of Gillespie syndrome. We consider Frizzled 4 gene could be related with both conditions. Frizzled 4 gene is expressed in cerebellar Purkinje cells, esophageal skeletal muscle and cochlear inner hair cells and the targeted deletion of this gene in rats exhibited distinct defects such as absence of a skeletal muscle sheath around the lower esophagus associated with progressive esophageal distension and dysfunction.[5]

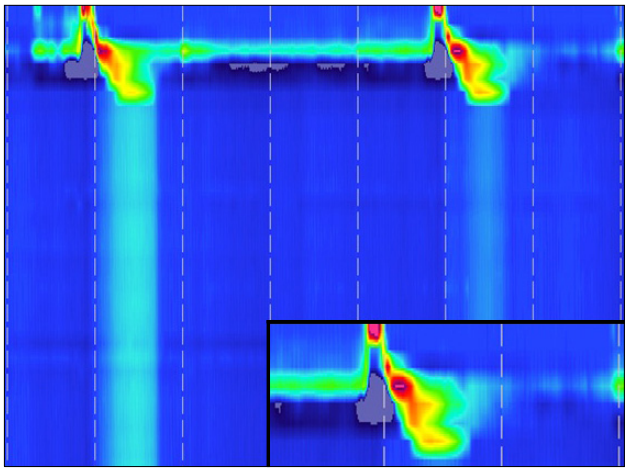

Figure. High-resolution manometry showing aperistalsis and a non-detected lower esophageal sphincter due to severe hypotonia.

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