

Joubert Syndrome among Two Related Children

Zakareya Alakri, MBBS, CABP* Ayman Khalil, MBBS, CABP**
Hakima Al Hashimi, FRCR, FFR RCSI, MHPE***

A seven-year-old Pakistani male presented with gross developmental delay. The child had delayed walking and speech. He had ataxic gait, head nodding to the left side and mild dysmorphic features.

Diagnosing Joubert syndrome (JS) requires clinical, radiological and genetic testing for final confirmation. Magnetic resonance imaging (MRI) study revealed Molar Tooth Sign (MTS) which is the hallmark, together with genetic testing, confirming the diagnosis of Joubert syndrome.

The manifestations of JS are variable and to keep into consideration the differential diagnoses of hypotonia and ataxia, delayed walking, speech and development.