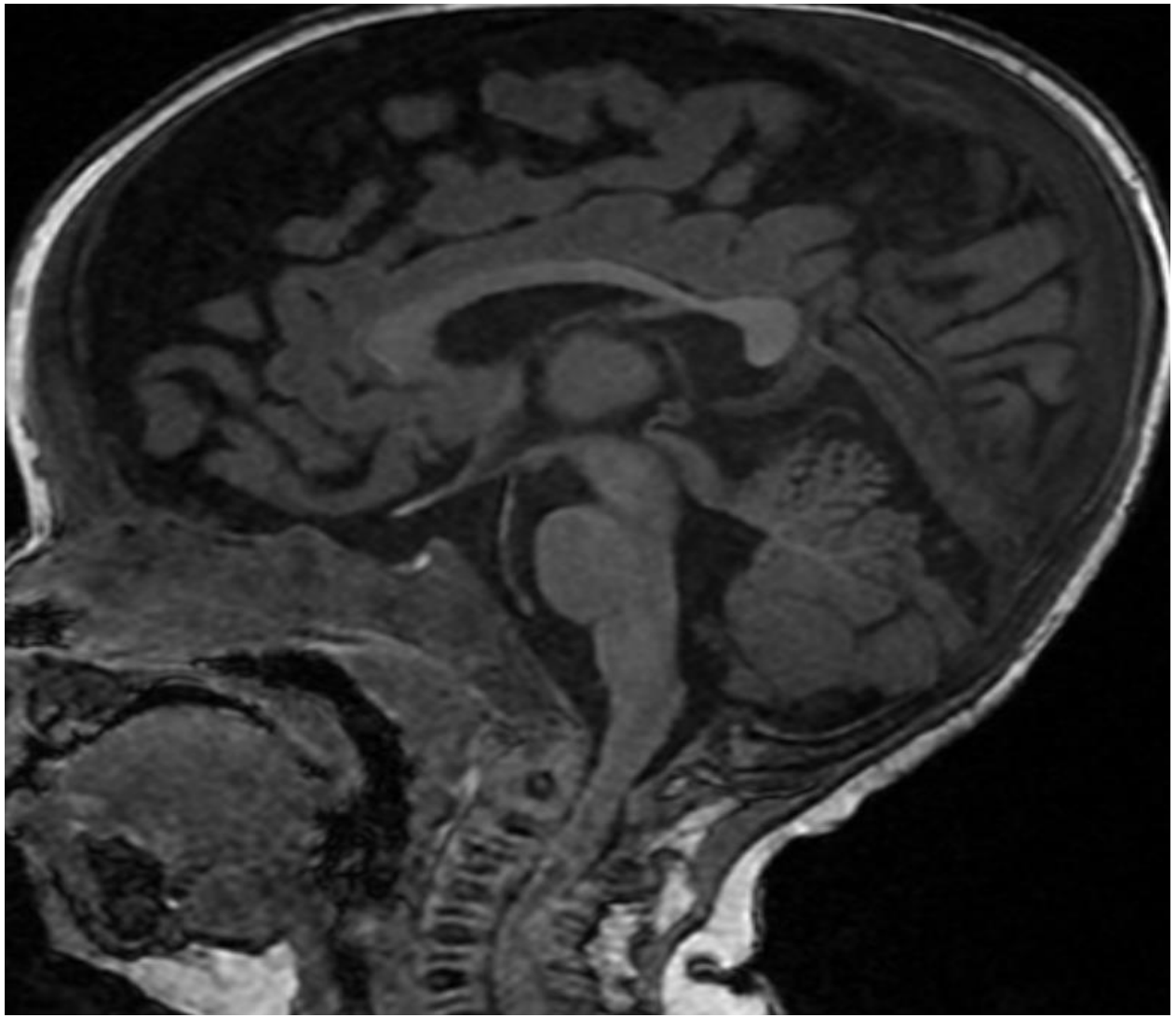
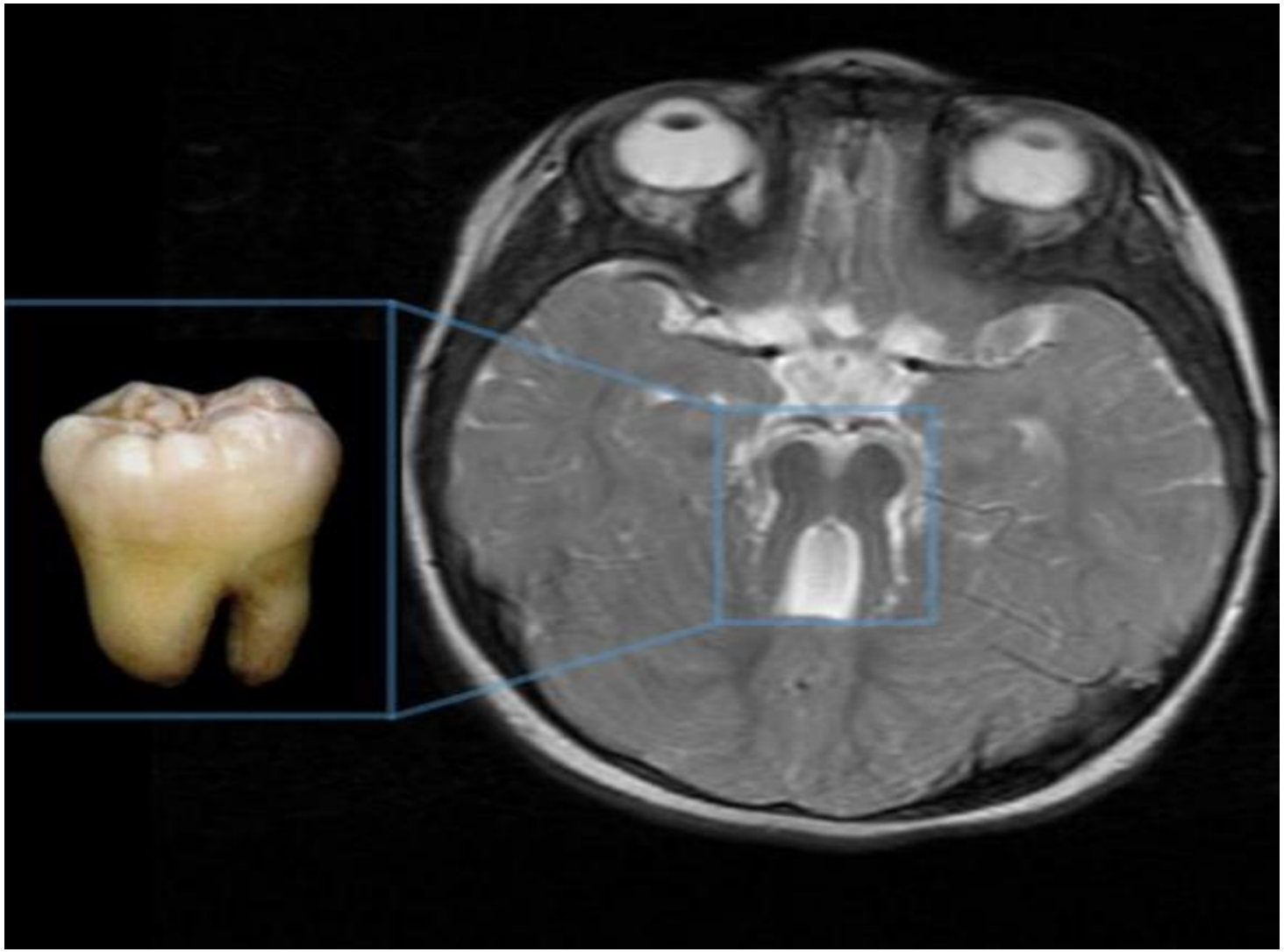
2025

KARNATAKA RADIOLOGY EDUCATION PROGRAM

CASE





MTS results from midlin**e hypoplasia of the cerebellar vermis, incomplete fusion of the halves of the vermis, abnormally deep interpeduncular fossa, and thick superior cerebellar peduncles .**

The batwing or umbrella sign can also be seen due to the **hypogenesis of the cerebellar vermis resulting in dilatation of the fourth ventricle**

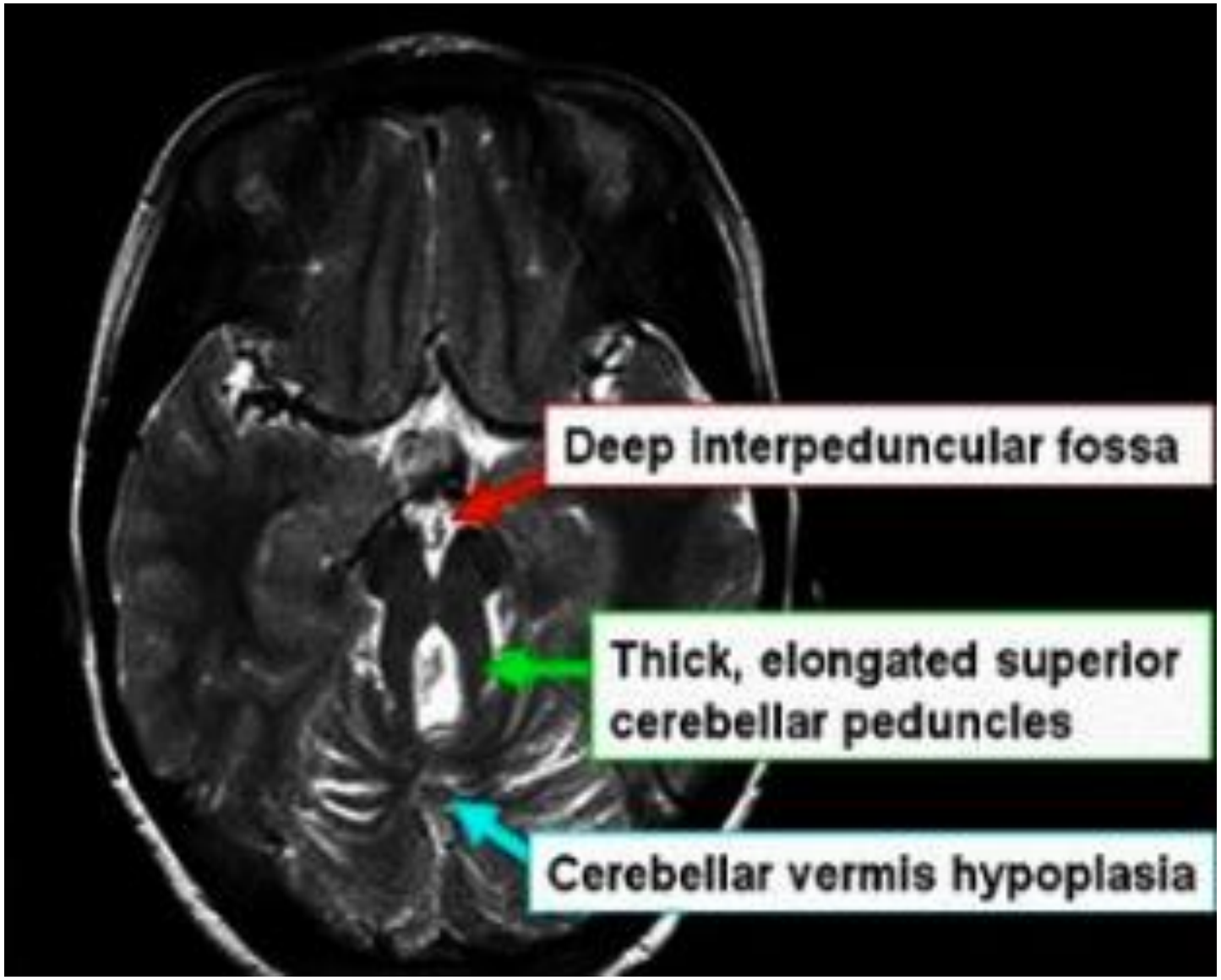
Histopathological studies have confirmed that the **gross appearance of the brainstem and cerebellum results due to the fragmentation of the dentate nucleus**

The pontomesencephalic junction is dysplastic with **abnormal decussation of the superior cerebellar peduncle.**

Another diagnostic feature noted on imaging is formed due to the **absence of the posterior verman lobe, leaving the cerebellar hemispheres separated by a cleft.**

Other findings include c**orpus callosum dysgenesis and moderate lateral ventricular enlargement.**

Clinical Subtype	Clinical Features
Pure JS	Hypotonia, ataxia, developmental delay, molar tooth sign
	No retinal or liver involvement
	No major gene associated with the phenotype
JS with ocular defect	Molar tooth sign, neurological features
	Retinal dystrophy and Leber's congenital amaurosis
	AHI1 gene most commonly mutated (20% of cases)
JS with renal defect	Molar tooth sign
	Nephronophthisis
	Absence of retinal involvement
	NPHP1 and RPGRIP1L genes commonly mutated
JS with hepatic defect	JS features
	Congenital hepatic fibrosis, chorioretinal or optic nerve colobomas, and nephronophthisis
	TMEM67 gene mutated in 70% of cases
JS with oculorenal defects	Neurological signs
	Retinal dystrophy, nephronophthisis
	CEP290 gene mutated in 50% of cases
JS with oro-facio-digital defects	Neurological features of JS
	Lobulated tongue, multiple oral frenula, mesoaxial polydactyly with y-shaped metacarpals, cleft lip/palate
	Hypothalamic hamartoma or congenital absence of the pituitary gland


Deep interpeduncular fossa
Thick, elongated superior cerebellar peduncles
Cerebellar vermis hypoplasia

Differential diagnosis

- Dandy-Walker malformation: the Dandy-Walker continuum may also occur simultaneously with the Joubert anomaly in some cases
- rhombencephalosynapsis
- mega cisterna magna: no structural abnormality