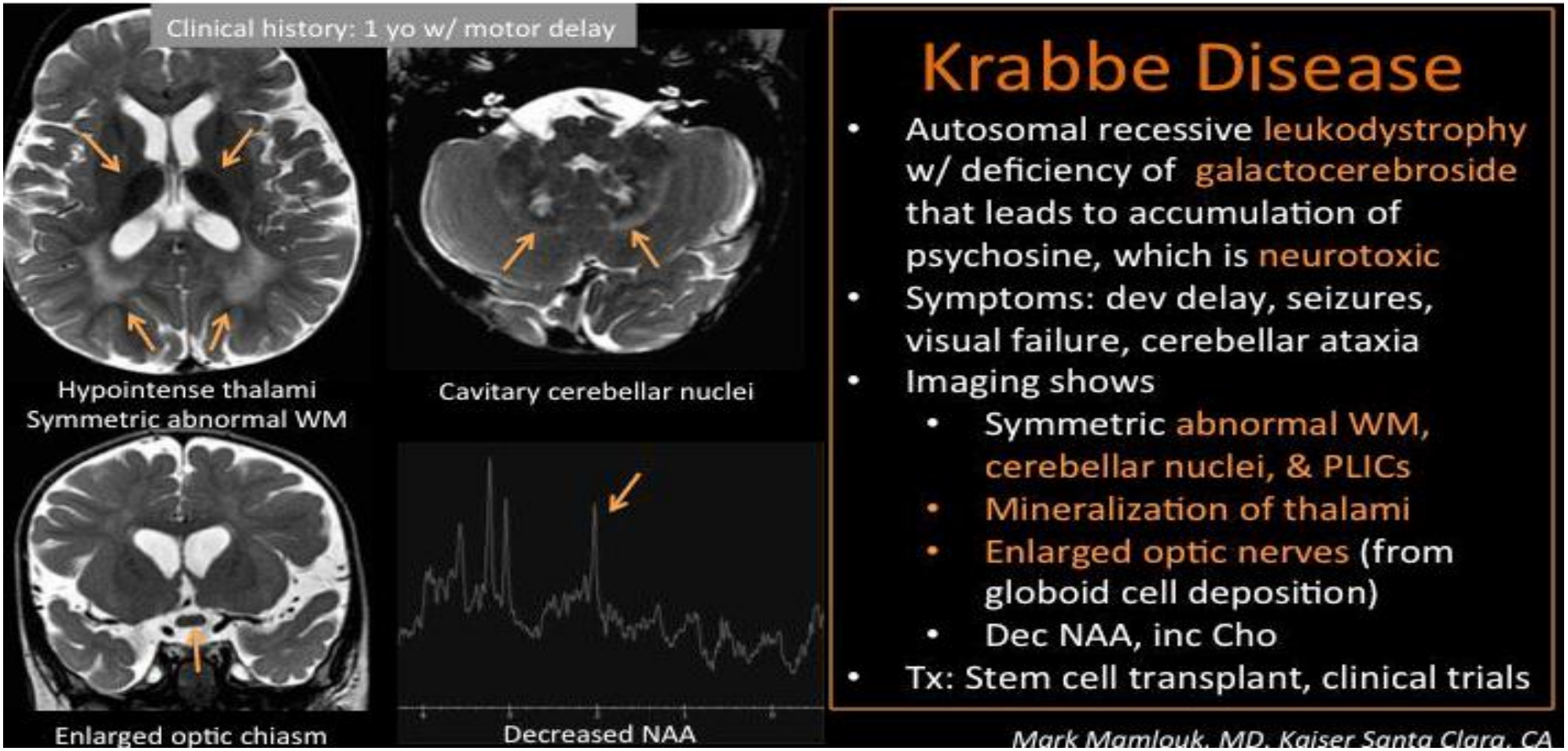
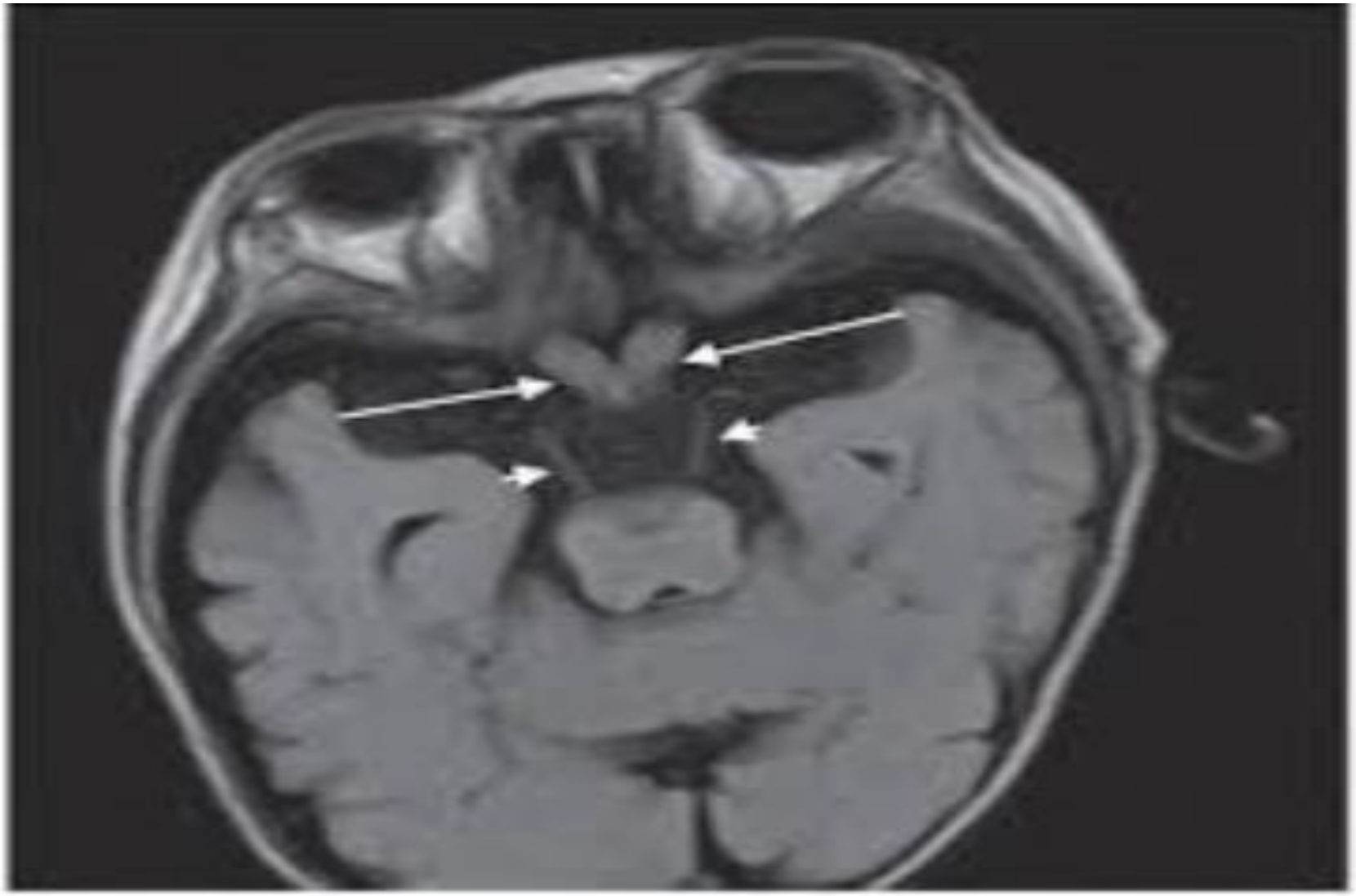
2025

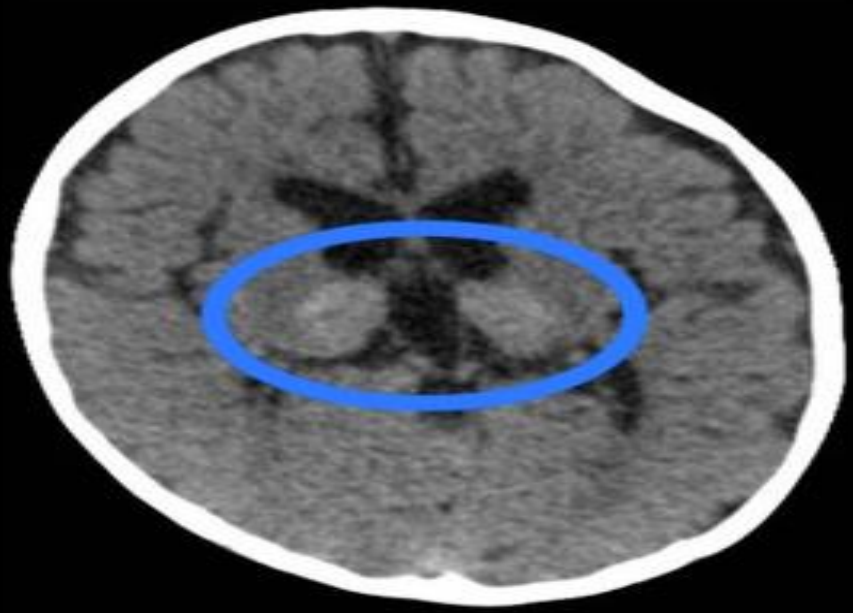
KARNATAKA RADIOLOGY EDUCATION PROGRAM

CASE


Clinical history: 1 yo w/ motor delay
Hypointense thalami
Symmetric abnormal WM
Cavitary cerebellar nuclei
Enlarged optic chiasm
Decreased NAA
Krabbe Disease
• Autosomal recessive leukodystrophy w/ deficiency of galactocerebroside that leads to accumulation of psychosine, which is neurotoxic
• Symptoms: dev delay, seizures, visual failure, cerebellar ataxia
• Imaging shows
  • Symmetric abnormal WM, cerebellar nuclei, & PLICs
  • Mineralization of thalami
  • Enlarged optic nerves (from globoid cell deposition)
  • Dec NAA, inc Cho
• Tx: Stem cell transplant, clinical trials
Mark Mamlouk, MD, Kaiser Santa Clara, CA



Krabbe Disease



- Krabbe disease is an autosomal recessive lysosomal storage disorder.
- There is an infantile form (85-90%), which occurs <2y age and is rapidly progressive. The adult form (10-15%) is more slowly progressive.
- The CT demonstrates bilateral hyperdense thalami and generalised cerebral atrophy.