A recent review of ISOD/MoCoD findings revealed that MRI initially shows extensive bilateral abnormal signal intensities suggesting edema of both gray and white matter with cytotoxic edema, and that edema subsequently decreases and curvilinear areas of reduced signal intensity appear at the gray/white matter junction, suggesting hemorrhagic deposits and laminar necrosis.
More than 90% of the cases present early, in the first few days of life with intractable seizures, hypertonia, and feeding difficulties.

The mean age at diagnosis was 12.5 months.

The median survival is 36 months.

The most common clinical findings are intractable seizures, feeding difficulties, dysmorphic facies.

Seizure is the most prominent initial sign and usually leads to a metabolic workup.
The clinical presentation of molybdenum cofactor deficiency, with or without sulfite oxidase deficiency, may mimic those of ISOD and Leigh syndrome. However, molybdenum cofactor deficiency may have a longer survival pattern.
The gross neuropathologic findings in brains of deceased patients with ISOD show severe atrophy of gyri, with cavitation of the centrum semiovale and increased ex vacuo ventricular enlargement.

- of CT and MR findings in ISOD describe cavitary white matter changes.
- In the absence of neonatal asphyxia, ISOD should be considered and pursued with further and appropriate biochemical testing
MR imaging performed at age 3 days. A, axial T2-weighted image; B, axial IR image; C, coronal T2-weighted image; D, sagittal T1-weighted image. There is abnormal high signal in the central and peripheral white matter in the T2-weighted images (A, C) and low signal in the inversion recovery T1-weighted image (B), with a cystic appearance of the white matter and a corresponding ex-vacuo enlargement of the ventricular system and subarachnoid spaces. Notice severe thinning of the corpus callosum (D, arrows) and flattening of the pontine protuberance.
Axial CT of the brain at the age of four days showing diffuse brain swelling causing effacement of cerebral sulci and compression of the lateral ventricles. White matter, basal ganglia, and thalami have remarkable low attenuation. B) Follow up at the age of three years and eight months revealed progressive white matter loss with associated enlargement of the lateral ventricles, cisterns, sulci, and fissures. Cysts are seen in the white matter and basal ganglia. Faint calcification is evident in both thalami.
Bilateral basal ganglia lesions:
1. Related to hypoxia
2. Carbon monoxide poisoning
3. Related to acquired metabolic disorders (hypoglycemia and kernicterus)
4. Related to inherited myelin disorders (canavan and Krabbe dis.)
5. Related to inherited metabolic disorders (leigh dis., Wilson, glutaric aciduria type I, methylmalonic acidemia, propionic aciduria, Cockayne disease, hypomyelination with atrophy of the basal ganglia and cerebellum, GM2 gangliosidosis)
6. Related to so-called degenerative disorders (Infantile bilateral striatal necrosis (IBSN))
7. Panthotenate kinase-associated neurodegeneration (PKAN) is an autosomal recessive disease
8. infections
Hypoxic–ischemic injury (acute asphyxia) in a term neonate
Leigh disease. Slightly asymmetrical involvement of the lentiform nuclei
Canavan disease. Typical involvement of the globi pallidi with sparing of the putamen and caudate
Infantile bilateral striatal necrosis
Wilson disease.
Huntington’s disease. Hyperintensity of the putamina and involvement of the caudate nuclei with widening of the frontal horns of the lateral ventricles
Panthotenate kinase-associated neurodegeneration. "Eye-of-the-tiger"
Fahr disease. Calcifications of the globi pallidi are hyperintense on T1WI and show no signal on SWI.
4-year-old-boy with Streptococcus sepsis. Basal ganglia are also affected with diffusion restriction: (a) DWI and (b) ADC map
Creutzfeldt–Jakob disease
Acute insults to the basal ganglia may sufficiently extensive to cause irreversible damage that can be detected radiologically. Although cystic encephalomalacia may be seen more commonly dystrophic calcification is present. Calcification is probably the most common abnormality of the basal ganglia seen in children. Although basal ganglia calcification may be ‘normal’ in adults, this is important and serves as marker of more extensive brain damage. Unfortunately, basal ganglia calcifications are not specific for any given disease and can be associated with a number of conditions.
THANK U