Maple Syrup Urine Disease (MSUD)

Why in News?

Scientists have created a new gene therapy for a debilitating genetic disorder called maple syrup urine disease (MSUD).

- **Recent Findings -** The treatment can prevent recurrence of deadly symptoms in a cow calf born with the disease.
- It prevented newborn death, normalized growth, restored coordinated expression of the affected genes and stabilized biomarkers in a calf as well as in mice.
- **Maple syrup urine disease (MSUD)** – It is a ***rare genetic disorder*** characterized by deficiency of an enzyme complex (branched-chain alpha-keto acid dehydrogenase).
- Branched-chain alpha-keto acid dehydrogenase is required to break down (metabolize) the 3 branched-chain amino acids (BCAAs) leucine, isoleucine and valine, in the body.
- The result of this metabolic failure is that all 3 BCAAs, along with a number of their toxic byproducts, (specifically their respective organic acids), all accumulate abnormally.
- In the classic, severe form of MSUD, plasma concentrations of the BCAAs begin to rise within a few hours of birth.
- If untreated, symptoms begin to emerge, often within the first 24-48 hours of life.
- **Types -** The classic type, intermediate type, intermittent type and possibly a thiamine-responsive type.
- **Cause** - When a mutated form of the BCKDHA, BCKDHB or DBT gene is inherited from both parents.
- **Symptoms -** Non-specific symptoms of increasing neurological dysfunction and include lethargy, irritability and poor feeding, soon followed by
    - Focal neurological signs such as abnormal movements, increasing spasticity and shortly thereafter, by seizures and deepening coma.
- It is characterized by a *distinctive sweet odor in the urine,* reminiscent of maple syrup.
- **Risk level -** If untreated, progressive brain damage is inevitable and death occurs usually within weeks or months.
- **Treatment** - The disorder can be successfully managed through a specialized diet in which the three BCAAs are rigorously controlled.
- However, even with treatment, patients of any age with MSUD remain at high risk for developing acute metabolic decompensation (metabolic crises).
    - Often triggered by infection, injury, failure to eat (fasting) or even by psychological stress.

Reference

The Hindu | Maple syrup urine disease



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