



Congenital Metabolic Diseases

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- $A \xrightarrow{\text{red X}} B$

- Accumulation of toxins
- Deficiency of essential products
- Defect in energy production or utilization
- Defect in cell structure



- CMD can present with different manifestations in every period of age.
- In neonatal period, CMD can present with asphyxia, sepsis, hydrops or neonatal jaundice.
- CMD, sometimes can present in intrauterine period



Screening of CMD

- In first few days of life in association of screening for hypothyroidism.
- Among several hundreds known types of CMD, only about 20 disorder are screened.
- Abnormal results of screening should be checked by confirmatory tests.



- Some of confirmatory tests, according to the suspected disorder:
- Serum Ammonia
- Serum Lactate
- Serum Homocystein
- Plasma aminoacids
- Plasma acylcarnitines
- Urine organic acids



Thank you for your attention