Galactosaemia in an infant: case report

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Abstract:

Galactosaemia is a disorder of galactose metabolism in which raised levels of galactose and galactose-l-phosphate damage various organs. It is a very rare disease (incidence 1 in 60,000) and the diagnosis is often missed, leading to poor prognosis. A case of clinical galactosaemia that was diagnosed at the age of 11 months is reported. It is important to be aware of this condition as early treatment may prevent some of the complications.