Severe neurological impairment in hereditary methaemoglobinaemia type 2.

Abstract: Recessive congenital methaemoglobinaemia (RCM) due to NADH-cytochrome b5 reductase (cytb5r) deficiency is a very rare disorder. We report on two unrelated patients (4 and 2.5 years old) with RCM type 2. Developmental delay was obvious at the age of 4 months. On follow-up, both children showed severe tetraspastic cerebral palsy, profound cognitive impairment, strabismus, impressive secondary microcephaly and failure to thrive. One novel mutation in the DIA1 gene was identified. Prenatal diagnosis was successfully done in both families by mutation analysis in chorionic villi or measurement of cytb5r in fetal amniotic cells. CONCLUSION: Due to the severity of this disease and its 25% recurrence risk, prenatal diagnosis should be made available to all affected families.

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