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Meckel Gruber syndrome: occurrence in non-consanguineous marriages.

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Source

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Abstract

Meckel Gruber syndrome is an uncommon, lethal, autosomal recessive disorder, associated consistently with polycystic kidneys, posterior encephalocele and polydactyly. We report three cases in non-consanguineous marriages, suggesting that the single gene defect occurs more commonly in non-consanguineous marriages than mutant genes associated with other autosomal recessive disorders that are usually related with consanguineous marriages. The usefulness of prenatal diagnosis is discussed.