

A new case of interstitial 1q 25.3-32.1 deletion: cytogenetic analysis molecular characterization and ultrasound findings.

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ABSTRACT

INTRODUCTION: deletion of long arm of chromosome 1(1q-) is a rare condition. Clinical features include Dwarfism, severe mental retardation, microcephaly and short neck delineating the "intermediate 1q deletion syndrome".

CASE REPORT: we report a new case of interstitial deletion of the long arm of chromosome 1, diagnosed in a 22+3 weeks gestation fetus in which cytogenetic analysis localized a loss of genetic materials of 18Mb in the 1q25.3-32.1. Fetal ultrasound showed neurodegenerative defects resembling Dandy-Walker's syndrome and bilateral clubfoot.

CONCLUSIONS: clinical characteristics of our case are markedly mild. This suggests that the type and the extension of the mutation obtained through cytogenetic studies, CGH array and ultrasound evaluation should be taken into account for prognostic evaluation and management of these patients.