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Seckel Syndrome with Spontaneous Chromosomal Instability

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Abstract: Seckel syndrome is an autosomal recessive disorder characterized by prenatal and postnatal growth retardation, bird-headed face and mild mental retardation. It is a disorder involving the DNA damage-response genes. Failure in the DNA damage response and repair process can cause chromosomal instability. In addition, it is possible that there are several loci responsible for this syndrome, and variety in the molecular pathogenesis is the cause of phenotypic heterogeneity. Three different loci have been reported thus far. The effect of the locus with mutation on phenotype may be used in the subgrouping of Seckel syndrome. We report a case with Seckel syndrome having spontaneous chromosomal instability. The patient had no hematologic or malignant disease although there was a severe chromosomal instability. To date, spontaneous chromosomal instability has been reported in two cases with Seckel syndrome.

Key Words: Seckel syndrome, chromosomal instability, chromosomal breakage syndrome

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