Three Male Cases with Isodicentric Y Chromosome Mosaicism Including 45,X Cell Line

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Objectives: 45,X/46,X,idic(Y)(q10) mosaicism with variable phenotypes is considered to be a rare sex chromosomal disorder. Here, we report three cases of isodicentric Y chromosomes with mosaic karyotype: a boy with short stature and two infertile male patients with non-obstructive azospermia.

Method: G and C banding, FISH and Y microdeletion techniques were performed using peripheral blood lymphocytes of the patients according to standard procedures.

Results: All three patients were detected to be mosaic as 45,X/46,X,idic(Y)(q10) and lacked the AZFb and AZFc regions on the Y chromosome.

Conclusion: 46,X,idic(Y)(q10)/45,X mosaicism may manifest in a spectrum of phenotypes. This variable phenotype that is noted in these patients are both due to the presence of isodicentric Y chromosomes and to the proportion and tissue distribution of specific cell lines as well, most notably 45,X. Importantly, isodicentric Yq with losses in the long arm of the Y chromosome may lead to the deletion of critical AZF region. In summary, detection of structural abnormalities of Y chromosome and mosaic pattern will provide necessary information allowing a better genetic counseling.

Key words: Mosaicism, infertility, short stature, isodicentric Y, microdeletion

Microcephalic Osteodysplastic Primordial Dwarfism Type Two

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Microcephalic osteodysplastic primordial dwarfism (MOPD) is a group of disorders similar to Seckel syndrome. Three subtypes have been described. We report the case of a 14.5-year-old girl who presented with intrauterine growth retardation, severe postnatal failure to thrive, facial dysmorphism, and skeletal dysplasia. The clinical and radiological findings were consistent with MOPD II. In addition to the previously published features, she also showed clinical and laboratory findings of severe insulin resistance. Our patient was the first child of the consanguineous Turkish parents. Her birth weight at term was 1.6 kg. Neuromotor development was mildly retarded. Physical examination revealed the following: height 102.5 cm [standard deviation score (SDS): -9.52], weight 29.6 kg (SDS: -3.6), head circumference 46 cm (SDS: -7), sitting height/height ratio 0.57; Tanner stage 5. She had a high forehead, prominent nose, retrognathia, small teeth, and high-pitched voice. Characteristic skeletal abnormalities were lumbar hyperlordosis, bilateral coxa vara, flaring of the distal femoral metaphysis, cranial displacement of the patellae, brachymetacarpia, and clinodactyly-V. She also had generalized obesity, acanthosis nigricans, and dry, thick skin with mild hirsutism. These clinical findings were consistent with insulin resistance. Her biochemistry was normal except for high triglyceride levels (333 mg/dL). Thyroid function tests revealed mild hypothyroidism. Surrenal function, insulin-like growth factor-1, and growth hormone stimulation tests were normal. Glucose was 191 mg/dL (10.5 mmol/L) and insulin was above 400 mU/mL at the 120th minute in the oral glucose tolerance test. Karyotype analysis was 46 XX. Pelvic and adrenal ultrasound showed no abnormalities. Cranial MR was normal. Direct sequencing of the coding region and the flanking intronic sequence of the exon 30 of the PCNT gene in the DNA sample of the patient revealed the homozygous mutations IVS30-2A>G. More case reports are needed to better delineate this rare type of MOPD.

Key words: Short stature, intrauterine growth retardation, facial dysmorphism, skeletal dysplasia, insulin resistance