



Low prevalence of glucokinase gene mutations in gestational diabetic patients with good glycemic control

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ABSTRACT. Glucokinase (GCK) plays a key role in glucose homeostasis. Gestational diabetes mellitus increases the risk of gestational complications in pregnant women and fetuses. We screened for mutations in coding and flanking regions of the *GCK* gene in pregnant women with or without gestational diabetes in a Brazilian population. A sample of 200 pregnant women classified as healthy (control, N = 100) or with gestational diabetes (N = 100) was analyzed for mutations in the *GCK* gene. All gestational diabetes mellitus patients had good glycemic control maintained by diet alone and no complications during pregnancy. Mutations were detected by single-strand conformation polymorphism and DNA sequencing. Thirteen of the 200 subjects had *GCK* gene

mutations. The mutations detected were in intron 3 (c.43331A>G, new), intron 6 (c.47702T>C, rs2268574), intron 9 (c.48935C>T, rs2908274), and exon 10 (c.49620G>A, rs13306388). None of these *GCK* mutations were found to be significantly associated with gestational diabetes mellitus. In summary, we report a low frequency of *GCK* mutations in a pregnant Brazilian population and describe a new intronic variation (c.43331A>G, intron 3). We conclude that mutations in *GCK* introns and in non-translatable regions of the *GCK* gene do not affect glycemic control and are not correlated with gestational diabetes mellitus.

Key words: Gestational diabetes; Glucokinase; *GCK*; SNPs; Mutations; Genetic susceptibility