Common genetic variations of imprinted genes that are maternally expressed could reveal the maternal-specific inheritance of birth weight. Human H19 is an imprinted growth regulatory gene located at chromosome 11p15.5 exclusively expressed from the maternal allele coding for a regulatory RNA implicated in fetal growth. To our knowledge H19 single nucleotide polymorphisms (SNPs) or their associations with birth weight have not been studied in Sri Lanka or in any native South Asian population. Here we report the association of maternal and newborn H19 rs2067051, rs217727 and rs2839703 genotypes on birth weight in a cohort of 196 healthy mother-baby pairs. Mothers had uncomplicated singleton pregnancies and the newborns were full term and healthy. The polymorphisms were studied using polymerase chain reaction (PCR) amplification and restriction fragment length polymorphism and confirmed by primer extension-based SNP analysis. The rarest allele and genotype respectively were T and TT for rs217727, A and AA for rs2067051 and G and GG for rs2839703 polymorphisms. Maternal H19 rs217727 TT genotype was associated with a significantly higher birth weight compared to CC genotype (one way ANOVA p<0.05; birth weight (mean+SD) 3.17+0.41, 2.93+0.46, 2.89+0.47 kg; in TT, CT and CC genotypes respectively). H19 rs2067051 genotype showed a statistically non significant increase in birth weight with the newborn A allele (one way ANOVA p>0.05, post test for linear trend p=0.022). H19 rs2839703 polymorphism was also not associated with birth weight. Thus maternal H19 rs217727 TT genotype is associated with a higher birth weight in healthy full term Sri Lankan newborns.

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