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Molecular Analyses of the *BORIS* Gene in Children with Silver-Russell Syndrome

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ABSTRACT Silver-Russell syndrome (SRS) is a heterogeneous disorder associated with intrauterine and postnatal growth retardation, skeletal asymmetry and facial dysmorphisms. In 7-10% of patients maternal uniparental disomy for chromosome 7 can be observed, nearly 50% of patients carry an epimutation resulting in hypomethylation of the imprinting center region 1 (ICR1) in 11p15. This leaves 40% of patients with unknown genetic aetiology. Based on the observation that the *CTCF* homologue *BORIS* is involved in imprinted gene expression and that it binds to methylated alleles we assumed that loss-of-function mutations in *BORIS* might have similar functional consequences as an ICR1 hypomethylation. On the other hand, there is evidence that *BORIS* mutations may disturb the methylation process and therefore cause hypomethylation in the ICR1. In our study we searched for *BORIS* gene mutations in a mixed cohort of SRS patients with and without 11p15 hypomethylation to determine whether this gene is involved in SRS aetiology. Mutation analyses revealed eight genomic variants but pathogenic mutations were not observed. Thus we conclude that alterations of the *BORIS* gene are probably not associated with SRS.