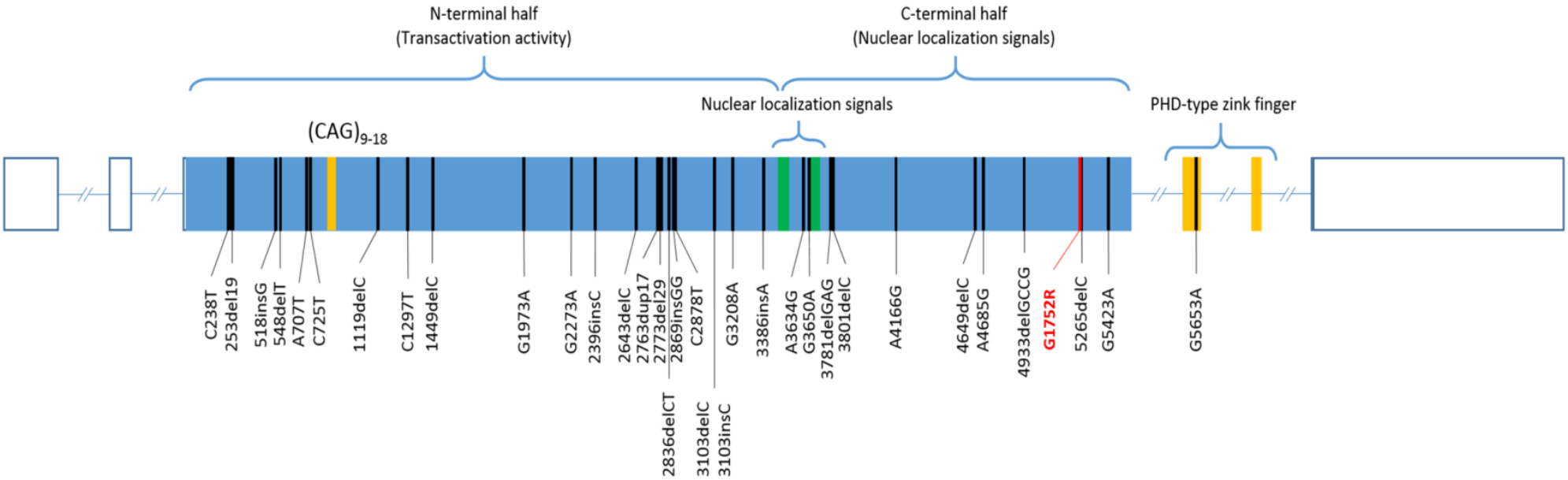

S2 Fig. Schematic representation of the *RAI1* gene with 6 exons and summarized point mutations found in SMS patients without deletion in 17p11.2.

Data on point *RAI1* mutations have been taken from appropriate papers [1-10]. The majority of reported *RAI1* mutations are located in exon 3 (blue box) containing ~ 98% of all *RAI1* gene coding sequence. Mutation c.5254G>A (p.Gly1752Arg [p.G1752R]) identified in present study is shown in red. $(CAG)_{9\text{-}18}$ – the range of CAG repeats earlier detected in SMS patients. Variability in the polymorphic CAG repeat length in the range of 10-16 repeats is common for general population [8] and apparently does not affect the SMS phenotype [1]. However, in study Bi *et al* (2006) was shown that combination of large $(CAG)_{18}$ repeat with c.3634A>G mutation in one patient could increase expression of the SMS phenotype [8].

References for S2 Fig

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