Mutations in \textit{WT1} in boys with sporadic isolated steroid-resistant nephrotic syndrome

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\textbf{ABSTRACT.} Mutations in the Wilms’ tumor gene, \textit{WT1}, can lead to syndromic steroid-resistant nephrotic syndrome and isolated steroid-resistant nephrotic syndrome. \textit{WT1} mutations have been identified in the majority of children with Denys-Drash or Frasier syndrome. \textit{WT1} mutations have not previously been identified in boys with sporadic isolated steroid-resistant nephrotic syndrome, but, recently, four boys with isolated nephrotic syndrome were identified to have \textit{WT1} mutations. However, whether boys with sporadic isolated steroid-resistant nephrotic syndrome should be routinely subjected to mutation analysis of \textit{WT1} has not been established. We examined 35 boys with sporadic isolated steroid-resistant nephrotic syndrome for mutations in \textit{WT1}. Mutation analysis of all 10 exons of \textit{WT1} was performed by polymerase chain reaction and direct sequencing. Karyotype analysis or Y chromosome identification was performed for all patients. A Y chromosome or a 46, XY karyotype was demonstrated for
all 35 patients. No causative WT1 mutation was identified in any of the patients. The WT1 mutation, IVS4+14T>C, which is not predicted to affect splicing, was identified in one patient who achieved complete remission after 8 weeks of oral prednisone treatment, indicating that IVS4+14T>C is not a causative mutation. Five WT1 polymorphisms were also identified in some patients and controls. Our results suggest that mutation analysis of WT1 should not be routinely performed for genetically defined boys with sporadic isolated steroid-resistant nephrotic syndrome.

**Key words:** Male; Mutation; Polymerase chain reaction; WT1; Steroid-resistant nephrotic syndrome