

## A Case of Denys-Drash Syndrome associated with Diaphragm Defect

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We report on a patient with male pseudohermaphroditism, glomerulopathy of which renal histology revealed diffuse mesangial sclerosis, and a diaphragm defect. A heterozygous missense mutation in the WT1 gene (366Arg to His) was identified and the patient was diagnosed as having Denys-Drash syndrome (DDS). In addition, his condition deteriorated to end-stage renal disease (ESRD) and peritoneal dialysis (PD) was begun at the age of 2 month. During PD, the child complained of recurrent pleural effusion on right side and had a diaphragm defect. The expression of the WT1 gene in pleural and abdominal mesothelium and the previous reports about the occurrence of diaphragmatic hernia in cases with WT1 mutations suggests that the diaphragm defect in this patient is part of the malformation pattern caused by WT1 mutations. In a case of DDS, nephropathy usually progressed to ESRD, and thereupon the PD is often applied to these patients. At that time, a checkup of diaphragm condition is necessary to prevent the complications such as pleural effusion.