Topic	先天性腎病症候群 **Congenital Nephrotic Syndrome**
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Nephrotic syndrome (NS) is defined by the presence of nephrotic-range proteinuria, edema, hyperlipidemia, and hypoalbuminemia. In children, nephrotic-range proteinuria is defined as protein excretion of more than 40 mg/m2/h or a first-morning urine protein/creatinine 2-3 mg/mg creatinine. According to the onset age of the disease, NS is classified arbitrarily as congenital NS (CNS, onset during 0-3 months-old), infantile NS (INS, onset during 3 months-old to 1 year-old) and childhood nephrotic syndrome (onset after 1 year old). CNS or INS can be found solitary without other anomalies, or with syndromic presentation such as Denys-Drash syndrome (caused by WT1 mutation) and Pierson syndrome (caused by LAMB2 mutaion). Secondary causes of CNS include perinatal infections of TORCH, Hepatits B, Hepatitis C, and HIV.

CNS of Finnish type (CNF) is a rare and severe kidney disorder starting soon after birth, characterized as premature birth, small for gestation age and enlarged placenta. Early onset nephrosis with heavy proteinuria, hyperlipidemia, hypercoagulopathy and immunocompromised status often caused poor growth and early mortality if not treated promptly. Nowadays, parenteral albumin supplement, hyper-caloric and protein abundant diet, medications for preventing and treating complications of NS have already improved the outcomes of the patients by making curative renal transplantation possible. In this presentation, we will describe a female newborn who was diagnosed as CNF incidentally before onset of the symptoms. Compound heterozygous mutation of NPHS1 gene was confirmed by exome-wide analysis of patient and her parents. NPHS1 gene encoded nephrin, which is the crucial protein for the structure of slit diaphragm. There is no definite relationship between type of mutation and prognosis of CNF. So treatment of CNS should be individualized in regard to whether to perform and timing of unilateral or bilateral nephrectomy.