Congenital nephrotic syndrome (CNS) of the Finnish type is an autosomal recessive disorder and caused by mutations in nephrin (NPHS1). It is the most common cause of CNS and is named because of its high incidence in Finland of 1:82000 live births.

A 31-year-old primigravida woman from Philippines was referred to our hospital at 17+1 weeks’ gestation with abnormally elevated maternal serum α-fetoprotein (MSAFP); 8.5MoM at 16+1 weeks’ gestation. Until then, the patient’s antenatal examination was unremarkable. MSAFP at our hospital was 11.246MoM. Acetylcholine esterase in amniotic fluid was negative and amniotic fluid α-fetoprotein was 41.09MoM.

Karyotype was normal. Neural tube defect and chromosomal abnormalities were excluded and CNS was suspicious. Analysis of the NPHS1 gene in amniotic fluid by massively parallel sequencing revealed a frameshifting deletion mutation in Exon 6(c.619del, p.Arg207Glyfs*28) and a missense mutation in Exon 9(c.1105C>T, p.Arg369Trp). Prenatal diagnosis was CNS.

Cesarean delivery was performed at 37+4 weeks’ gestation due to transverse presentation. A live baby girl weighing 2730g was delivered. The accessory placenta was noted, however, weight of placenta was 500g, which is not placentomegaly. The neonate was not edematous and she urinated well without proteinuria. Creatinine, blood urea nitrogen level and electrolytes in serum were normal. Laboratory studies to diagnose infections; toxoplasma, rubella, cytomegalovirus, and herpes simplex virus were normal.

CNS is expressed in 40-80% of cases with the above genetic mutations, and the symptoms usually appear within 2 weeks to 1 month of age. Therefore, close monitoring including blood and urine tests will be performed regularly. Treatment of CNS is generally supportive. When children are nearing end-stage kidney disease or if the nephrotic syndrome cannot be managed by medical means, bilateral nephrectomies and kidney transplantation performed, usually between 1-3 years of age.

**Fig. 1.** Fetal karyotype analysis with amniocentesis, The fetal karyotype was normal

**Fig. 2.** Sequencing chromatogram of the CNS proband. Sequence analysis of the proband and parents demonstrated c.619del (A) and c.1105C>T (B) of NPHS1 gene.