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# Disease modeling of Karyomegalic Interstitial Nephritis using patient derived induced pluripotent stem cells

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**Objectives:** Karyomegalic interstitial nephritis (KIN) is a very rare disorder caused by the mutation in FAN1 gene on 15q13.3 and manifested by karyomegaly and gradual progress of fibrosis in target organ such as kidney, liver and brain. We diagnosed KIN in 52 year old woman. Kidney biopsy showed typical kayromegaly in tubule cells and FAN1 gene sequencing showed the homogenous p.Gly663Ilefs*54 mutations.

**Methods:**

Peripheral blood mononuclear cells were reprogrammed using non-integrative viral transduction.

**Results:** Kidney biopsy showed typical kayromegaly in tubule cells and FAN1 gene sequencing showed the homogenous p.Gly663Ilefs*54 mutations. We generated an induced pluripotent stem cell (iPSC) line (iPSC-KIN) using peripheral mononuclear cells isolated from her and same p.Gly663Ilefs*54 mutation was detected DNA isolated from iPSC-KIN cell line. We re-differentiate this iPSC-KIN into proximal renal tubular epithelial cells and it showed distinct karyomegaly as well in electromicroscopy.

**Conclusions:** This iPSC-KIN model may serve as a disease model for KIN biopsy and also as innovative therapeutic tool for the treatment of hereditary kidney disease like KIN.