KÁLMÁN TORY

Semmelweis University
Faculty of Medicine
1st Department of Paediatrics

Address: Bókay János u. 53-54., H-1083 Budapest, Hungary

RESEARCH AREA

The research group formerly identified the first variant with a trans-associated mutation-dependent pathogenicity in an autosomal recessive disorder (NPHS2 R229Q) [Tory et al, Nat Genet, 2014) and a novel gene of steroid-resistant nephrotic syndrome (DKC1). They demonstrated the role of rRNA-pseudouridylation in DKC1-associated nephrotic syndrome [Balogh et al, PNAS, 2020]. The group created a novel population-genetic algorithm to identify novel incompletely penetrant variants and interallelic interactions in autosomal recessive disorders [Mikó et al, Hum Mutat, 2021]. Function and interallelic interactions of the most frequently implicated protein in steroid-resistant nephrotic syndrome, podocin, as well as the function of novel genes are studied in cell culture experiments and on a Caenorhabditis elegans model.

TECHNIQUES AVAILABLE IN THE LAB

Cell culture experiments, vector construction, mutagenesis, PCR, rtPCR, qPCR, sequencing, expression, immunostaining, characterization of protein localization, maintenance, transformation, mutagenesis of Caenorhabditis elegans strains, fluorescence stereomicroscope, population-genetic calculations.

SELECTED PUBLICATIONS

Mikó, Á., Kaposi, A., Schnabel, K., Seidl, D., **Tory, K.** (2021) Identification of incompletely penetrant variants and interallelic interactions in autosomal recessive disorders by a population-genetic approach. **Hum Mutat 42:** 1473-87.

Balogh, E., Chandler, J.C., Varga, M., Tahoun, M.K., Menyhárd, D., Schay, G., Goncalves, T., Hamar, R., Légrádi, R., Szekeres, Á., Gribouval, O., Kleta, R., Stanescu, H., Bockenhauer, D., Kerti, A., Williams, H., Kinsler, V., Di, W.L., Curtis, D., Kolatsi-Joannou, M., Hammid, H., Szőcs, A., Perczel, K., Maka, E., Toldi, G., Sava, F., Arrondel, C., Kardos, M., Fintha, A., Hossain, A., D'Arco, F., Kaliakatsos, M., Koeglmeier, J., Mifsud, W., Mooseja, M., Faro, A., Jávorszky, E., Rudas, G.H., Saied, M., Marzouk, S., Kelen, K., Götze, J., Reusz, G., Tulassay, T., Dragon, F., Mollet, G., Motameny, S., Thiele, H., Dorval, G., Nürnberg, P., Perczel, A., Szabó, A.J., Long, D.A., Tomita, K., Antignac, C., Waters, A.M., **Tory, K.** (2020) Pseudouridylation defect due to DKC1 and NOP10 mutations cause nephrotic syndrome with cataracts, hearing impairment and enterocolitis. **Proc Natl Acad Sci USA 117:** 15137-47.

Mikó, Á.K., Menyhárd, D., Kaposi, A., Antignac, C., T**ory, K.** (2018) The mutation-dependent pathogenicity of NPHS2 R229Q: a guide for clinical assessment. **Hum Mutat 39:** 1854-60.

Stráner, P., Balogh, E., Schay, G., Arrondel, C., Mikó, Á., L'Auné, G., Benmerah, A., Perczel, A.K., Menyhárd, D., Antignac, C., Mollet, G., **Tory, K.** (2018) C-terminal oligomerization of podocin mediates interallelic interactions. **Biochim Biophys Acta Mol Basis Dis. 1864:** 2448-2457.

Tory, K., Menyhard, D.K., Woerner, S., Nevo, F., Gribouval, O., Kerti, A., Straner, P., Arrondel, C., Cong, E.H., Tulassay, T., Mollet, G., Perczel, A., Antignac, C. (2014) Mutation-dependent recessive inheritance of NPHS2-associated steroid-resistant nephrotic syndrome. **Nat Genet 46:** 299-304.