

TAMÁS SZABÓ



National Academy of Scientist Education, 3rd year

Semmelweis University
Faculty of Medicine, 3rd year

YEAR OF BIRTH

2003

FORMER SZENT-GYÖRGYI PUPIL

no

RESEARCH UNIT

Semmelweis University

SZENT-GYÖRGYI MENTOR

Kálmán Tory

JUNIOR MENTOR

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SPECIALIZATION

pediatrics,
genetics,
nephrology,
molecular biology

SECONDARY SCHOOL

Piarist High School in
Kecskemét

NAME OF TEACHER

Ildikó Kiss

LANGUAGES

English/advanced

IMPORTANCE, AIMS AND POSSIBLE OUTCOME OF RESEARCH

Alport syndrome is a genetic disorder which is caused by a variant occurring in the genes of type IV collagen (*COL4A3*, *COL4A4*, *COL4A5*). The inheritance of the disorder is really complex. *COL4A3* and *COL4A4* is located on chromosome 2 while *COL4A5* is located on chromosome X. There is also a chance of dominant inheritance in the case of autosomal chromosomes. The laboratory previously published an algorithm (Mikó et al, Hum Mutat, 2021), which we use for the observation of the disorder. We would like to identify variants with incomplete penetrance. If there are such variants we would like to search interallelic interactions. Among the heterozygote variants we would like to search signs of dominant negative effect.

AMBITIONS AND CAREER GOALS

During the university I would like to get as much knowledge as I can which I think will be useful in my future career. The fact that I joined to a research group is helping me in this goal. I suppose that I gain way of thinking here which can help me solve various kinds of problems in my future.

HONORS AND PRIZES

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PUBLICATIONS

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