Boston Children's Hospital

HARVARD MEDICAL SCHOOL TEACHING HOSPITAL

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Post-doctoral Fellow
Project: Discovery and modeling of genetic forms of childhood kidney disease

Contact information:
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Ranked #1 Pediatric Nephrology by *U.S.News & World Report*
Ranked #1 Children's Hospital by *U.S.News & World Report*

Laboratory Description: I am faculty at Boston Children's Hospital and Harvard Medical School. My research explores the genetic basis of pediatric kidney diseases with a focus on Mendelian genetic forms of nephrotic syndrome and kidney stone disease. I employ a combination of cell biology, proteomics, and mouse models to explore the pathways that regulate nephrotic syndrome in these genetic forms. I am funded through an NIH K Career Development Grant, additional foundation grants, and a startup package from Boston Children's Hospital (see CV attached).

Position and Project Descriptions: Full-time post-doctoral fellow to begin **Summer 2021** for 2-year commitment. In the first project, the fellow would perform cell biology and mouse biology experiments to explore *TRIM8* variants as a cause of nephrotic syndrome in children (see TRIM8 publication below). Our group is actively developing CRISPR cell and mouse models of this disease to understand the pathogenic mechanisms by which patient variants cause disease and develop biological assays for future therapeutic studies. Research responsibilities would include (i) investigating cellular dysfunction caused by *TRIM8* variants in knock-in cells, (ii) exploring what protein interactions and enzymatic functions of TRIM8 are altered by these variants, (iii) modeling TRIM8 variants in a CRISPR-based knock-in mouse model. In a second project, we are beginning a two-year study at Children's where we will be recruiting and performing genomic analyses in subjects with early onset kidney stone disease. Research responsibilities would include (i) conducting chart review and data collection of newly recruited patients, (ii) receiving training in and performing exome and other genomic analyses, (iii) participating in review of clinical genetics findings with our clinical nephrology team. The ideal candidate would have significant experience in wet laboratory research, and I would train them in genomics.

Please check out these publications:
https://experiencejournal.childrenshospital.org/directory/physicians/m/amar-majmundar
https://connects.catalyst.harvard.edu/Profiles/display/Person/118690
https://advances.sciencemag.org/content/7/1/eabe1386/tab-article-info
https://www.cell.com/ajhg/fulltext/S0002-9297(21)00008-2
https://www.kidney-international.org/article/S0085-2538(17)30494-5/abstract