

Human Genetics Unit

(Sri Lanka)

Brief description of the “Clinical Center” or “Services on undiagnosed rare diseases”

Location – Human Genetics Unit, No. 25, Kynsey Road, Colombo 08, Sri Lanka.

Contact person – Prof. Vajira H.W Dissanayake, Chair professor of Anatomy and Director, Human Genetics Unit, Faculty of Medicine, University of Colombo.

Research staff – Prof. Vajira H. W. Dissanayake –Chair professor and Director, Human Genetics Unit, Faculty of Medicine, University of Colombo. Dr. Dineshani Hettiarachchi – Lecturer, Department of Anatomy, Faculty of Medicine, University of Colombo. Dr. Nirmala Sirisena – Senior Lecturer and Clinical Geneticist, Department of Anatomy, Faculty of Medicine, University of Colombo. Dr. Kalum Wetthasinghe – Senior Lecturer and Medical Geneticist, Department of Anatomy, Faculty of Medicine, University of Colombo. Dr. Hemali W. W Goonasekara – Senior Lecturer and Consultant Haematologist, Department of Anatomy, Faculty of Medicine, University of Colombo. Nilaksha Netthikumara – Bioinformatician, Human Genetics Unit, Faculty

Past (six months) and future/upcoming events related to undiagnosed rare disease. Please indicate date and venue.

International conference on birth defects- November 2019. We would like to invite UNDI to have their next meeting during this time as a satellite meeting of the conference.

Relevant scientific publications (selected and / or published by the center)

The first Sri Lankan family with Dent disease-1 due to a pathogenic variant in the CLCN5 gene: a case report. Ranawaka R, Sirisena ND, Dayasiri KC, Cogal AG, Lieske JC, Gamage MP, Dissanayake VHW. BMC Res Notes. 2017, 30;10(1):539.

Novel AGXT gene mutation in a Sri Lankan family with primary Hyperoxaluria type 1. Sirisena ND, Tantrigoda NH, Kariyawasam KJ, Jayasekara RW, Dissanayake VH. Nephrology (Carlton). 2016;21(1):75-6.

Novel COL4A3 gene mutations in a consanguineous family with autosomal recessive Alport syndrome. Sirisena ND, Thalgahagoda S, Abeyagunawardena A, Neumann M, Schudlach HO, Jayasekara RW, Dissanayake VH. Nephrology (Carlton). 2015;20(8):580. Erratum in: Nephrology (Carlton). 2016 Apr;21(4):346.

Grants. a) grant award to announce funding for projects; b) Received grants from other organizations/institutions for undiagnosed rare diseases research.

University Grants Commission Sri Lanka 4.2 million Sri Lankan rupees (25,000 USD)

Contact person

Prof. Vajira H.W Dissanayake, Chair professor of Anatomy and Director, Human Genetics Unit, Faculty of Medicine, University of Colombo.

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