

Guest lecture: New Born Screening for inborn errors of Metabolism

Speaker: Dr Inderneel Sahai, Associate Prof., University of Massachusetts, Worcester, Harvard Medical School, Boston, MA

Department of Medical Education, AIIMS, Rishikesh scheduled a Guest Lecture on 20 Nov 2017 delivered by Guest Speaker Dr Inderneel Sahai, Associate Prof., University of Massachusetts, Worcester, Harvard Medical School, Boston, MA. Inderneel Sahai, is a practicing Clinical Geneticist in Boston, MA. Dr. Sahai graduated from Armed Forces Medical College in 1987 and has been in practice for many years. In her very illuminating talk, Dr. Inderneel Sahai stressed on how genetic disorders are now recognized to play an increasing important role in pediatrics. She explained that close to 10% of diseases among hospitalized children have been ascribed to Mendelian traits inherited as single gene defects, not a surprising figure considering that approximately 1000 inborn errors of metabolism (IEM) have been identified to date, primarily through the detection of endogenous metabolites abnormally accumulated in biological fluids and tissues. She elaborated that while dramatic advances in molecular genetics have greatly changed the landscape of diagnostic options for many genetic disorders, a biochemical approach remains the dominant force for the diagnosis and monitoring of IEMs.

She also stressed on public awareness to initiate at-risk screening (prenatal diagnosis), newborn screening (testing of pre symptomatic patients) and high-risk screening (testing of symptomatic patients) in India. Session was chaired by Dr Soprakash Basu, Professor, Department of Surgery, AIIMS Rishikesh.



