

## **Students' Annual Seminar**

# **Structure and dynamics of a cataract-active mutant G57W of human GammaS Crystallin**

## **Khandekar Jishan Bari**

Cataract, or opacification of the eye lens, is the leading cause of blindness world over. While age-related cataract is the result of the accumulation of environmental and metabolic effects, congenital cataract, seen in newborn children, is essentially genetic in origin (1,2). A mutant of human  $\gamma$ S-crystallin, G57W, has been recently reported in a Chinese family wherein a young boy and his mother were found to have cataract in the center of the lens (3).

We attempt to structurally characterize the G57W mutant and study its dynamics by solution NMR. The 3D structural characterization, dynamics and biophysical studies of this mutant along with its individual domains and its comparison with the wild type human  $\gamma$ S-crystallin will be discussed.

### **References:**

1. Sun H, Ma Z, Li Y, Liu B, Li Z, Ding X, Gao Y, Ma W, Tang X, Li X, Shen Y (2005) Gamma-S crystallin gene (CRYGS) mutation causes dominant progressive cortical cataract in humans. *J Med Genet* 42(9):706–710
2. Brubaker WD, Freites JA, Golchert KJ, Shapiro RA, Morikis V, Tobias DJ, Martin RW (2011) Separating instability from aggregation propensity in gammaS-crystallin variants. *Biophys J* 100(2):498–506
3. Khan I, Chandani S, Balasubramanian D (2016) Structural study of the G57W mutant of human gamma-S-crystallin, associated with congenital cataract. *Mol Vision* 22:771–782

***Friday, Apr 20<sup>th</sup> 2018***

***03:30 PM (Tea/Coffee at 03:00 PM)***

***Seminar Hall, TIFR-H***