Haemoglobinopathies Satellite Centre, Chandrapur

Sickle Cell Clinic, and a Cohort Study on Sickle Cell Disease (SCD) Patients

Sickle Cell Clinic

- ✓ The Clinic started in association with Civil Hospitals in **Chandrapur and Gadchiroli** districts in January 2016
- ✓ The Rural and Sub-District Hospitals at **Mul, Sindewahi, Warora, and Wani** are visited, new patients are enroled and followed up every three months time interval
- ✓ Activities being done through the clinic:
  - Clinical check-up
  - Molecular confirmation of the disease, and CBC, biochemical analysis (KFT, LFT, HCV, HBsAg, Ferritin)
  - Tab. Folic acid is given to patients
  - Regular monitoring of SCD patients on Hydroxyurea (HU) therapy
  - X-ray and USG are done for the patients as and when required

Cohort study

Aim: To explore the hematological, molecular and clinical features of SCD patients longitudinally.

Cases, 749 [SS:625, Sβ:118, SD:6] → ***Enrolment & Methods*** → HPLC, CRDB, ARMS-PCR, CBC, LFT, KFT, Ferritin

- The two most common α-globin gene deletions in Indian population (α/3.7 and α/4.2) were determined in 329 SCA, and 65 Sickle/β-thalassemia patients.

Results:

- ✓ Among the patients (**Table 1**), majority of them were HbS Homozygous followed by Sickle/β-thalassemia 118 (14.1%), and 8 (0.96%) patients had compound heterozygosity conditions with HbS and other mutations in β-globin and γ-globin chains.
- ✓ Also, among Sickle/β-thalassemia patients, 114 (96.6%) of them had HbS+IVS 1-5(G-->C), 3 (2.5%) had HbS+CD15(G-->A) and one patient had HbS+CD8/9(+G) β-globin chain mutations.
- ✓ A total of 388 SCA and 52 Sickle/β-thalassemia patients were on HU therapy (10-15 mg/Kg/day).
- ✓ Patients with SCA, and Sickle/β-thalassemia on HU therapy coming regular to follow-up showed decrease episodes of painful crisis, requirement of blood transfusion and hospital admission than those of without HU **(Fig 1a & 1b)**.

Table 1: SCD & compound heterozygotes among the cases

Condition	No. cases (%)
SCA	655 (78.8)
Sickle/β-thalassemia	118 (14.1)
β-thalassemia major	48 (6.0)
HbS +HbD Punjab	6 (0.7)
HPFH+HbS	1 (0.1)
HbS + δβ-thalassemia	1 (0.1)
δβ-thalassemia+IVS1-5(G→C)	1 (0.1)
HbE+ IVS 1-5 (G-->C)	1 (0.1)
Total	831

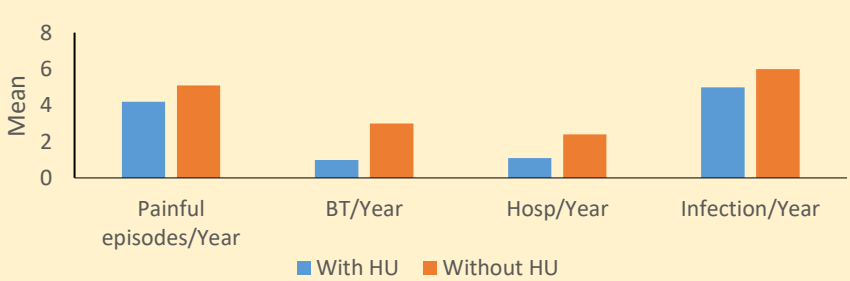

Fig. 1a: Clinical findings of SCA patients taking Hydroxyurea (HU) at 3, 6, 9, 12, 15; 18 months' time interval

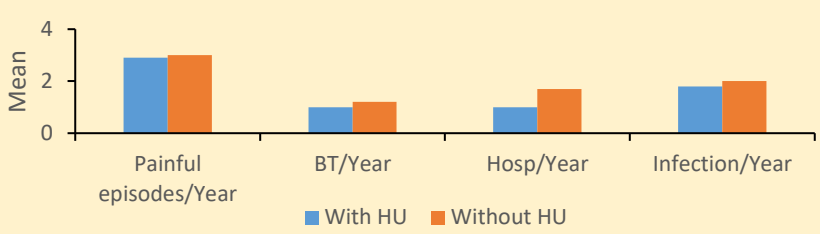

Fig. 1b: Clinical findings of Sickle/β thalassemia patients taking HU at 3, 6, 9, 12, months' time interval

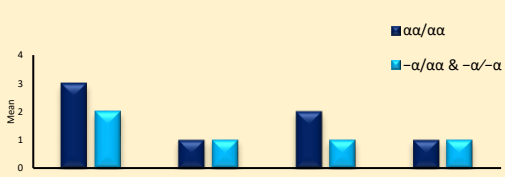

Fig. 2a: Clinical profile of SCA cases with alpha deletion and with normal α-allele

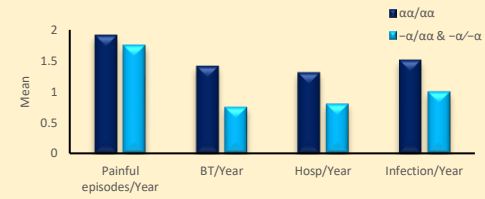

Fig. 2b: Clinical profile of Sickle/β-thal cases with alpha deletion and with normal α-allele

- ✓ Among the patients with SCA and Sickle/β-thal, normal α-globin genotype (αα/αα), was found in 283 (84.3%) and 53 (85.4%) respectively.
- ✓ Homozygosity for α-globin deletion (-α/-α) was noted to be higher 2 (0.6%) among SCA patients than Sickle/β-thal patients 2 (3.7%).
- ✓ Patients with α-globin gene deletion had lesser episodes of painful crisis, infections, hospitalization, and blood transfusion requirement **(Fig 2a & 2b)**.

Conclusion: Findings of the present study suggest that identification of genetic modifiers and longitudinal assessment with hematological parameters could be helpful in the management of Sickle Cell Disease.