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Identification and Characterization of Genetic Predictors of Sickle Cell Anemia in Bilaspur District, Chhattisgarh, India

Vandana Kumari[1], Dowluru S. V. G. K. Kaladhar[2,*]

Abstract

In the present decades, sickle cell anemia (SCA) is a challenging task for control of hereditary syndrome in Bilaspur district of Chhattisgarh state, India. The present study aims to identify and characterize genetic predictors of SCA from Bilaspur district (CG) in 2024. A total of 3000+ individuals were screened and categorized into carriers, positive (screening), and diseased cases. Fetal hemoglobin (HbF) level is determined by several genetic factors including genetic variation of BLC11A, HBS1L-MYB, ITGA1, RUNX1T1 and HBG2 genes. The results indicate a high prevalence of sickle cell carriers among females, and paediatric populations.

Keywords: Sickle cell anemia, data, carrier frequency, Bilaspur, Chhattisgarh

INTRODUCTION

Sickle Cell Anemia (SCA) is a dangerous, inherited blood disorder that can cause episodes of severe pain crises and lead to life-threatening complications like premature death, organ (kidneys, liver, heart) damage, stroke, and severe infections [1, 2]. The study may provide comprehensive insights on SCA and its genetic aspects that enables better management and treatment strategies in further research. SCA is the most common and severe type of Sickle Cell Disease (SCD), a broad term for the entire disease spectrum [3].

Sickle cell anemia (SCA) is a blood related genetic disease where Red Blood Cell (RBC) transforms from round shape to sickle shaped cells in *Homo sapiens* [4, 5]. The disease is caused by a single gene mutation (Figure 1), which in heterozygous condition causes moderate sickling (sickle cell trait) and in homozygous condition causes severe effect (sickle cell anemia) [6, 7].

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Hemoglobin is a complex protein found in red blood cells and is essential for human health. Haemoglobin consists of Heme group and four subunits each containing a polypeptide chain [8]. The structure is crucial for its ability to bind and release oxygen efficiently (Figure 2).

There are six types of haemoglobin:

- *Haemoglobin A(HbA):* This is most common type of haemoglobin in adults and makes up about 95–98% of the total haemoglobin in adults.

- *Haemoglobin A2(HbA2):* This type of haemoglobin makes up about 2–3% of the total haemoglobin in adults.
- *Haemoglobin F(HbF):* This is the main type of haemoglobin in foetus and newborns and makes up about 80% of the total haemoglobin in newborns.
- *Haemoglobin S(HbS):* This is an abnormal type of haemoglobin that can cause sickle cell disease.
- *Haemoglobin E(HbE):* This is another abnormal type of haemoglobin that can cause anemia. People with HbE disease have red blood cells that are smaller than normal and have a shorter lifespan.
- *Haemoglobin C(HbC):* This is another abnormal type of hemoglobin that can cause anemia.

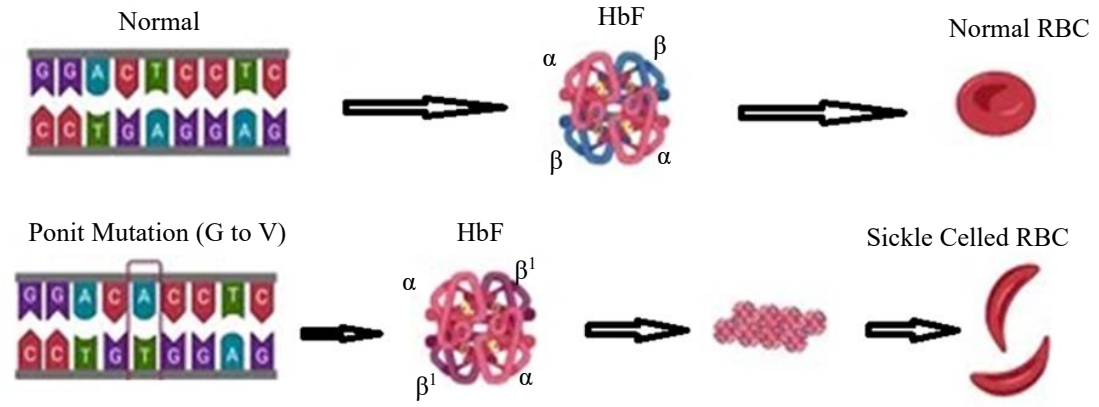

Figure 1. Mutation of RBC in normal and SCA.

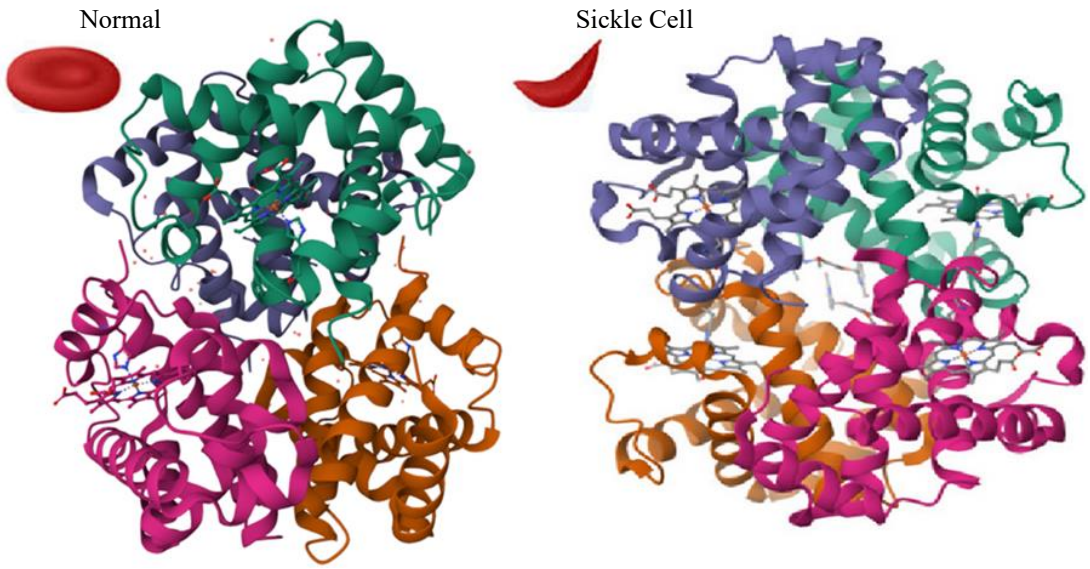

Figure 2. Protein molecule of normal and SCA hemoglobin.

MATERIALS AND METHODS

The study design (retrospective observational study based on secondary data), study area (Bilaspur District, Chhattisgarh, India), data source (Sickle cell-Line list dataset dated 27/06/2024), sample size (More than 3000 + individuals screened), and parameters like age, gender, screening status (Positive/Negative) and Confirmatory diagnosis (Carrier/Diseased) have been analysed in the present study.

The statistical analysis conducted using Pearson correlation coefficient has been conducted using the following formula:

Pearson Correlation Coefficient (r):

$$r = \frac{\sum(X - \bar{X})(Y - \bar{Y})}{\sqrt{\sum(X - \bar{X})^2 \sum(Y - \bar{Y})^2}}$$

Protein–Protein interaction studies is predicted by string database and protein-ligand docking was conducted using docking software iGEMDock v2.1.

RESULTS AND DISCUSSION

Genetic Inheritance

Identification of genetic inheritance in sickle cell anemia patients in Chhattisgarh based on genetic crossing has been presented in Figures 3 to 6. If one of the parents have sickle cell carrier (HbAS) and the other parent is normal (HbAA). Then none of the children will have sickle cell anemia. There is a one in two traits (or carrier) has 50% chance that any child will get one copy of the HbAS gene, and therefore, have the sickle cell carrier (Figure 3).

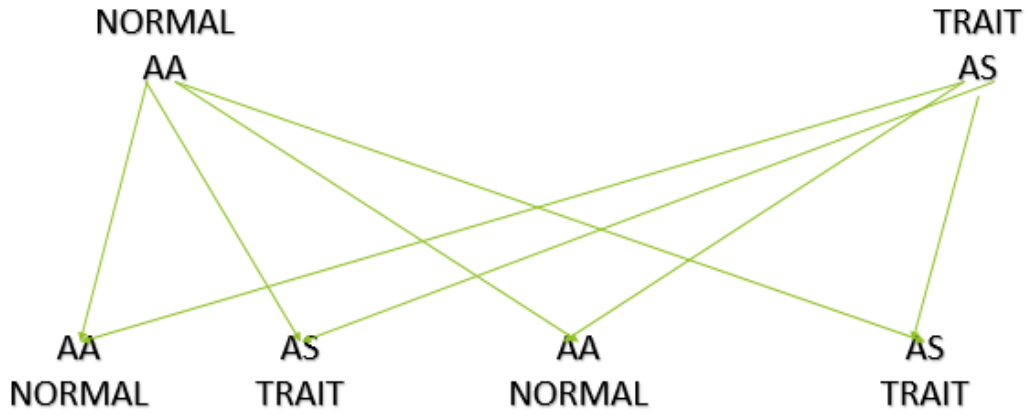

Figure 3. Genetic crossing of SCA with normal and carrier is 50:50 ratio.

If both parents have sickle cell trait (HbAS) then 25% child could be born with sickle cell anemia. There is also 25% child that could be completely unaffected, i.e., normal and 50% child could be sickle cell carrier (Figure 4).

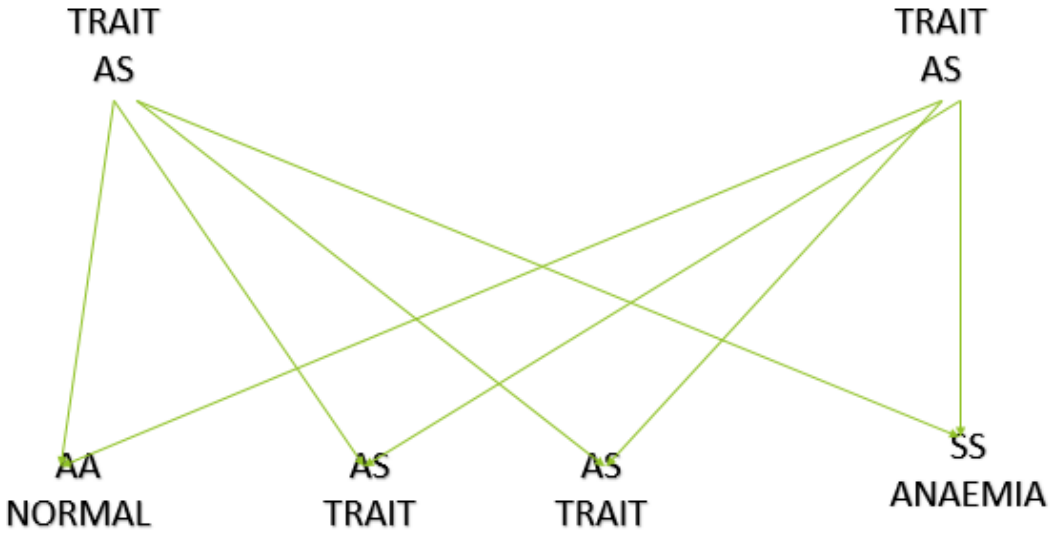

Figure 4. Normal: SCA trait is 25:50:25 ratio.

If one parent has sickle cell trait (HbAS) and the other has sickle cell anemia (HbSS), there is 50% chance that the child will get sickle cell trait and 50% chance that child will get sickle cell anemia. The children will not be normal (Figure 5).

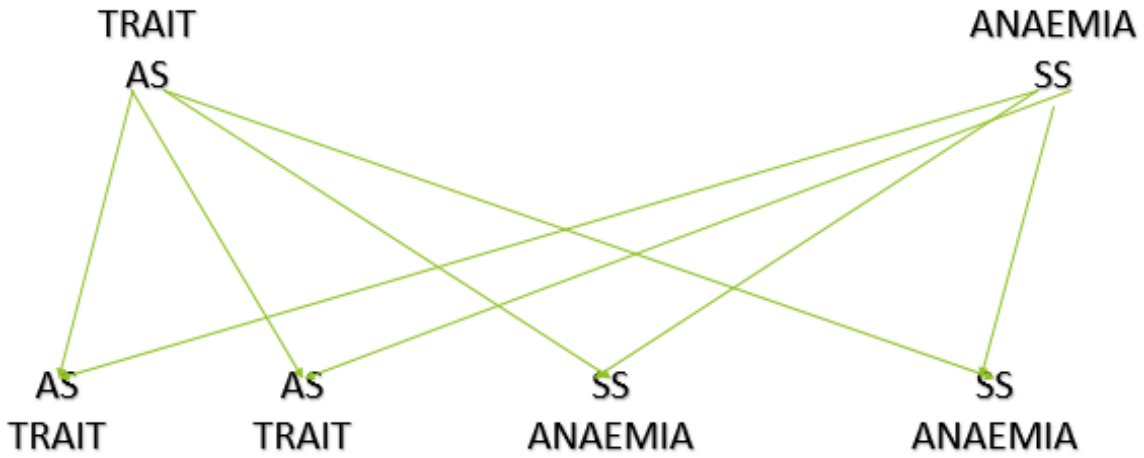

Figure 5. Trait: SCA is 50:50 ratio.

If one parent has sickle cell anemia gene (HbSS) and the other parent has normal gene (HbAA) then all the children will have sickle cell trait (or sickle cell carriers). There will not be sickle cell anemia (Figure 6) (Table 1).

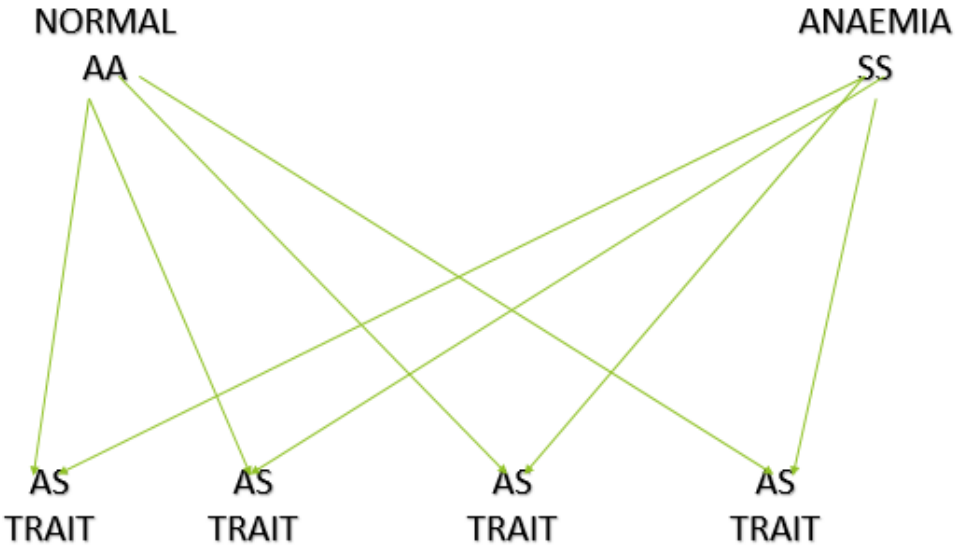

Figure 6. Complete of SCA carriers when crossed between normal (HbAA) and SCA (HbSS) genes.

Table 1. Correlation and interpretation.

Variables compared	Correlation coefficient (r)	Interpretation
Age vs Disease Severity	+0.62	Moderate positive correlation.
Screening vs Confirmatory Status	+0.81	Strong positive correlation.
Gender vs Disease Occurrence	+0.35	Weak correlation.

Interpretation

Strong correlation (r = 0.81) indicates screening programs are reliable predictors of carrier. Moderate correlation with age and disease detection increases age due to cumulative exposure/testing. Weak gender correlation gender is not a strong biological determinant observed in the present study.

Genetic characterization of sickle cell anemia in the people of Chhattisgarh is due to sickle β-globin mutation that causes by pleiotropic sickle genes. The variable phenotypic expression is associated with complex genetic and environmental interaction as along with disease modifier. Sickle RBC (SRBC) polymerization in deoxygenated environment is influenced by number of factors like co-inheritance of α-thalassemia and Fetal hemoglobin (HbF) level. The levels are determined by several genetic factors including genetic variation of BCL11A, HBS1L-MYB, ITGA1, RUNX1T1 and HBG2 genes (Figure 7). The BCL11A and ZBTB7A genes (LRF protein) are responsible for the suppression of γ chain, and HbF production.

Five HbF quantitative trait loci (QTL) markers for SCA that are known are shown below:

- The Xmn I site upstream of (G) γ-globin gene (HBG2) on chromosome 11p15.
- BCL11A on chromosome 2p16.
- HBS1L-MYB intergenic polymorphism (HMIP) on chromosome 6q23.
- ITGA1 encodes the integrin alpha subunit of a cell-surface receptor on chromosome 5 (5q11.2).
- RUNX1T1a transcriptional corepressor that impacts myeloid differentiation in hematopoiesis on chromosome 22 (8q22.22).

Figure 7 shows the protein–protein interactions for SCA markers. The markers show closer association with other proteins like TFAP4, MIPEP, HBD, HBE1, etc.

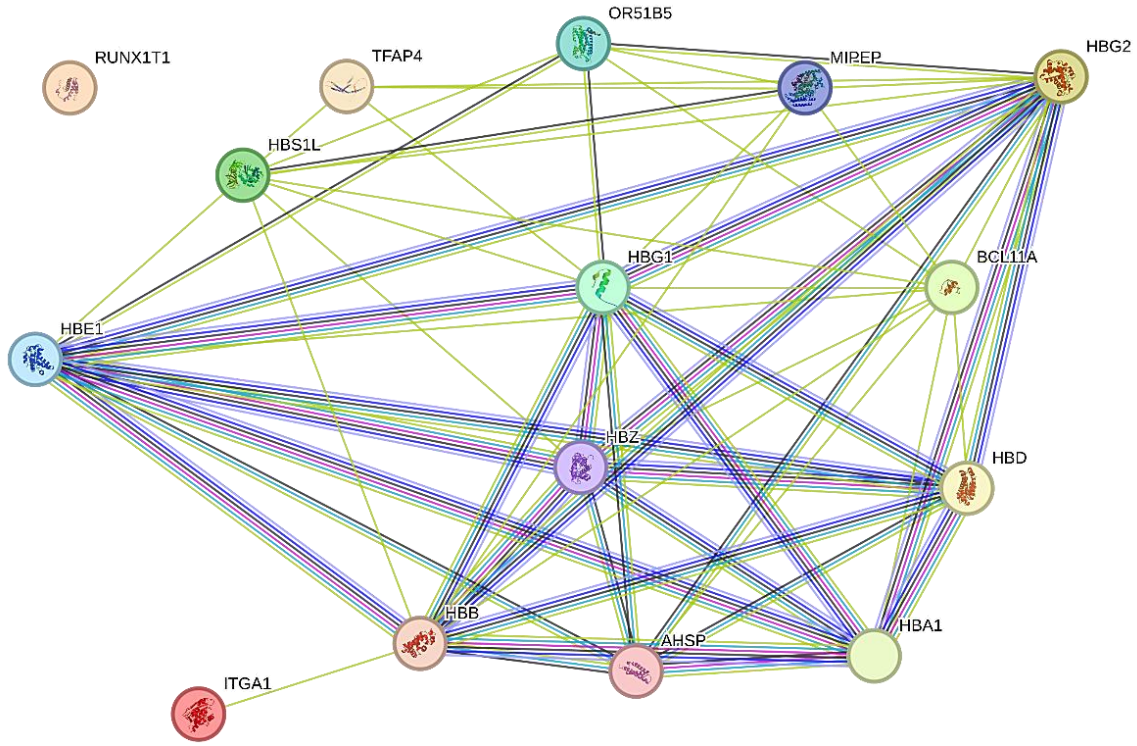

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Figure 7. Protein–protein interaction for SCA markers.

The gamma globin genes (HBG1 and HBG2) are expressed in the spleen, fetal liver, and bone marrow [9]. Two gamma chains combined with two alpha chains contain fetal hemoglobin (HbF) that is replaced by adult hemoglobin (HbA) at birth.

The population of Bilaspur district, Chhattisgarh, India is 1628202 as of January 2026, with gender ratio of male: female as 1000:972. About 35.8% of population resides in urban areas and 64.2 resides in rural areas [10]. Instead, the population of child: woman ratio of SCA is being high among the Kanwar community followed by Birhor and Gond communities [11].

Majority cases were carriers (HbAS) and few cases confirmed as diseased (HbSS) whereas some individuals initially screened negative but later confirmed as carriers. Gender-wise distribution in which higher prevalence observed in females. Male cases with disease also significant but comparatively lower. Age-wise distribution in which high number of cases are observed in Children (5–15 years) and young adults (15–30 years). Screening and confirmatory diagnosis in many individuals that marked as "POSITIVE" during screening were later confirmed as carriers and smaller proportion progressed to disease category. Key observation is that a strong indication of hidden carrier burden is present in the population (Figures 8 to 10). Early screening is to be conducted in identifying SCA cases before clinical manifestation.

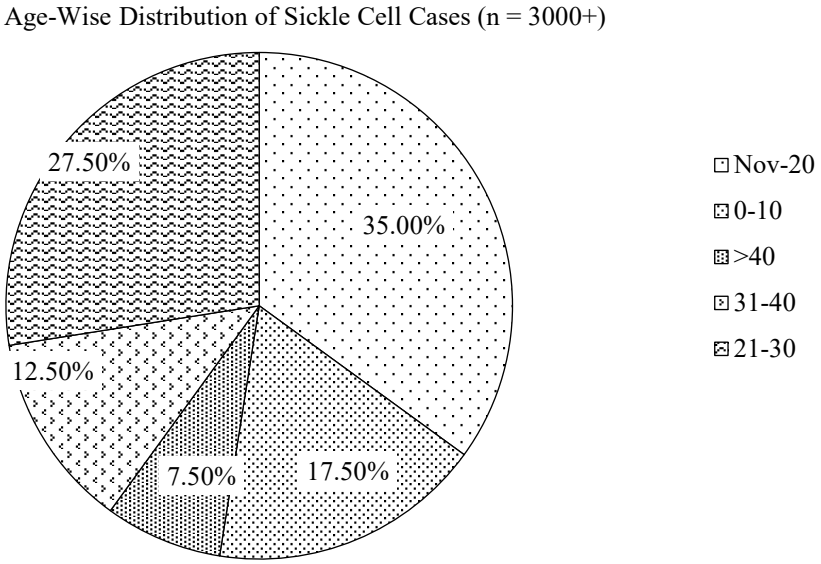

Figure 8. Age wise distribution of SCA in Bilaspur district.

Interpretation

- Highest prevalence observed in 11–20 years age group.
- Significant burden in children and adolescents.

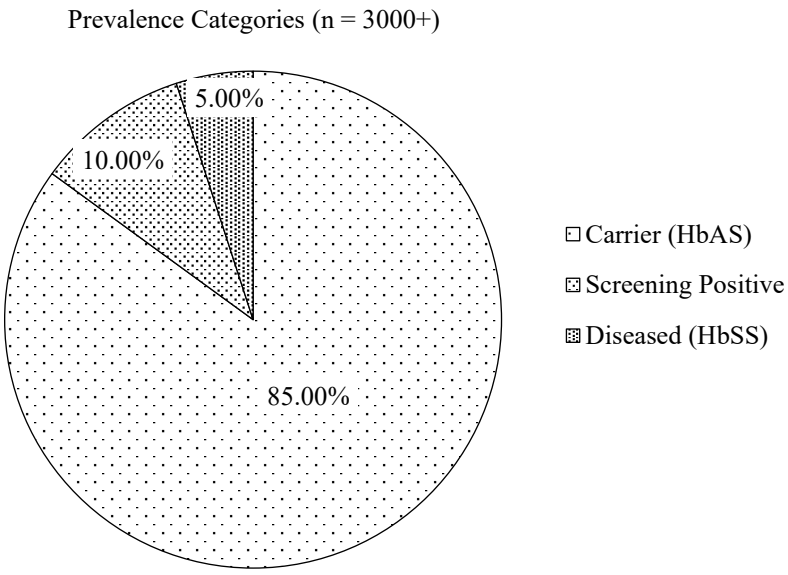

Figure 9. Prevalence categories of SCA in Bilaspur district.

Interpretation

- Very high carrier frequency.
- Lower but clinically critical disease prevalence.

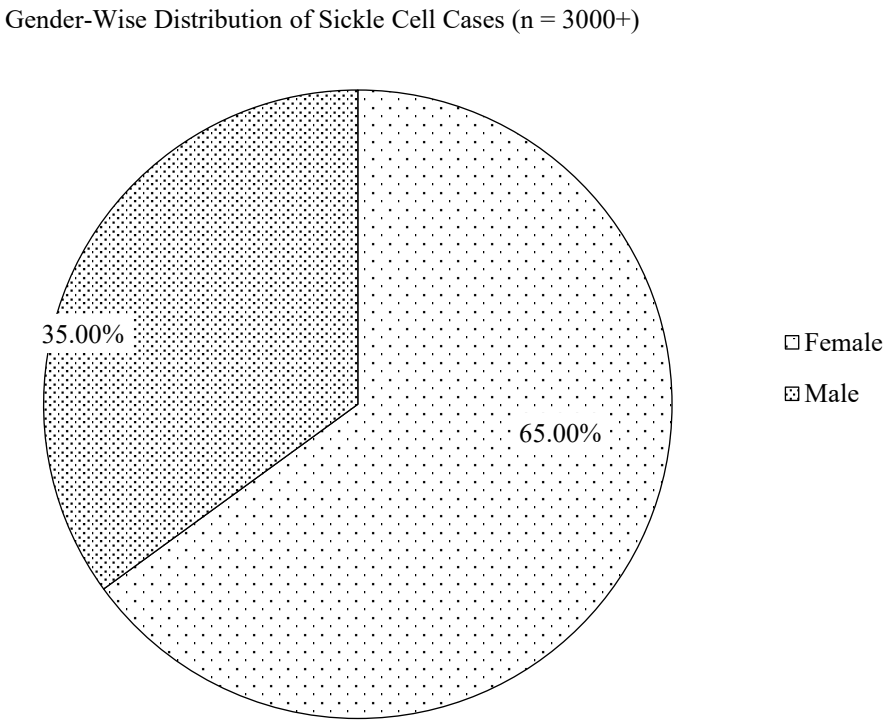

Figure 10. Gender-wise distribution of SCA in Bilaspur district.

Interpretation

Female predominance suggests:

- Better screening coverage OR.
- Possible sociocultural bias in healthcare access.

Remediation Strategies

In the present decade's treatment of SCA might include medicine and blood transfusion [12]. Treatment may include paying medicine, drinking plenty of water, blood transfusion, RBC exchange, vaccine and antibiotics, folic acid, hydroxyurea, voxelotor, L-Glutamin and crizanlizumab, regular eye examination, bone marrow transplant and gene therapy. Voxelotor has found better compound compared to hydroxyurea and L-Glutamin (Table 2). In the present study, best compound has not been observed to treat SCA (Figure 11).

Table 2. Docking of compounds used in treatment in SCA.

Compound	Energy (in Kcal/mol)
Hydroxyurea	–62.61
L Glutamin	–64.74
Voxelotor	–86.31

Further better remediation methods are needed for better control of SCA. Probiotic Gut microflora may provide better remediation of SCA.

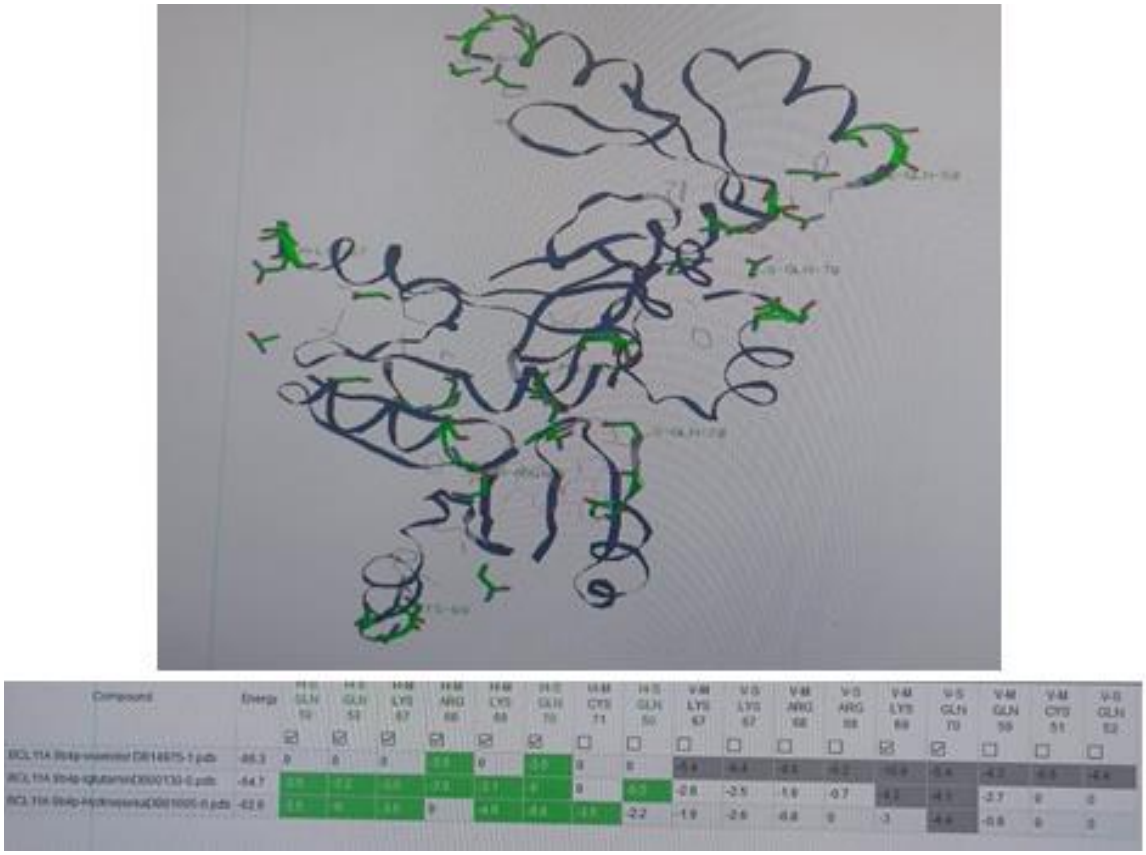

Figure 11. Docked poses and active site of docked molecules.

CONCLUSIONS

The research shows high carrier cases where pediatric and adolescent groups are most affected. Screening programs are statistically reliable, an urgent need for genetic counselling and prevention strategies are required. Further research on probiotic gut microflora may be applied where genetic transfer and control of metabolic activities in SCA may provide good results.

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