Differential Diagnosis:

- Griscelli syndrome
- Hermansky-Pudlak syndrome

Final Diagnosis: CHEDIAK HIGASHI SYNDROME

Management

- Treatment of infections with Trimethoprim-sulfamethoxazole 5mg/kg/day into two divided doses until HSCT is available.
- Supportive care with blood and platelet transfusions
- Definitive treatment: Allogeneic hematopoietic stem cell transplantation (HSCT)

CONCLUSION

A four-year-old child born full term out of a non-consanguineous marriage is brought with complaints of recurrent fever and skin infection since childhood. The child was born with light colour and skin and appears pale on presentation.

After clinically evaluating the child and running laboratory investigations and genetic testing with LYST gene mutation, a diagnosis of **CHEDIAK HIGASHI SYNDROME** was made and symptomatically treated with antibiotics, until the mainstay treatment option- hematopoietic stem cell transplantation is possible.

Regular follow-up is advised to monitor progression to the accelerated phase.