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Prevalence of sickle cell disease and sickle cell trait among children below 17 years of age attending Entebbe regional referral hospital in Wakiso district. A cross-sectional study.

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Abstract.

Background:
Sickle cell disease (SCD) is a genetic disorder that affects the shape of hemoglobin in the red blood cell, leading to the formation of a sickle shape. This study aims to determine the Prevalence of sickle cell disease and sickle cell trait among children below 17 years of age attending Entebbe Regional Hospital, Wakiso district, Uganda.

Methods:
This was a prospective cross-sectional study conducted at Entebbe regional referral hospital, which included 100 respondents who were selected using a purposive and convenience sampling technique, and data were collected prospectively by administering researcher-guided questionnaires.

Results:
The study participants, 55% (55/100), were female, and the age range of respondents was 0– 17 years, 45.0% (45/100) were aged 6-11 years. The Prevalence of sickle cell disease and sickle cell trait among respondents was 19.0% (19/100), with 6% (6/100) respondents found having the homozygous form, HbSS, and 13% (13/100) found having the heterozygous form, HbAS. 86% (86/100) of the respondents had heard about sickle cell, and the majority, 92% (92/100) of the parents had children that had never been screened for sickle cell, with only 8% (8/100) of the children ever screened. 39% (39/100) of the parents reported knowing the cause of sickle cell disease, with the majority, 61% (61/100), not knowing how SCD is caused. 65% (65/100) of the parents of respondents reported that sickle cell is transmitted from parent to child, 17% (17/100) did not know how SCD is transmitted.

Conclusions:
The prevalence of sickle cell disease and sickle cell trait was high, while the majority of the participants lacked knowledge about sickle cell screening.

Recommendations:
There is a need for health facilities to scale up screening services for sickle cell disease in the community so as to enable early diagnosis and treatment of the disease and reduce morbidity and mortality.

Keywords: *Sickle cell disease, sickle cell screening, sickle cell trait, children below 17 years, Entebbe Regional Hospital.*
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Background of the study
Sickle cell disease (SCD) is an inherited blood disorder caused by mutations in the hemoglobin subunit beta (HBB) gene and is associated with premature mortality and significant morbidity, including vasoocclusive pain, stroke, and multi-organ damage. The protective association between variants in HBB and severe Plasmodium falciparum malaria results in a higher prevalence of HBB mutations in geographic areas with high malaria prevalence (CDC, 2024).

Sickle cell disease (SCD) remains a major neglected tropical disease in Africa. The disease is a genetic disorder that

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affects the shape of haemoglobin in the red blood cell, leading to the formation of a sickle shape. The sickle shape contributes to co-morbidities throughout the lifetime, including pulmonary hypertension, stroke, organ damage, gall bladder disease, and premature mortality. Globally, 250,000 out of 300,000 infants born with haemoglobin disorders every year have SCD. Furthermore, an estimated 240,000 children in low and middle-income countries are born with SCD annually, of which 50%-80% die before the age of five years (Asiimwea *et al.,* 2023).

Worldwide, sickle haemoglobinopathies lead to a substantial burden of disease that is not adequately addressed. Accurate data are lacking, but the worldwide estimate for neonates born with sickle cell disease each year is 400 000, including 300 000 with sickle cell anaemia. The greatest burden is seen in sub-Saharan Africa, where more than 75% of all sickle cell disease occurs, with this proportion projected to increase by 2050. In Africa, sickle cell disease contributes substantially to mortality in children younger than 5 years and, therefore, limits progress towards achieving UN Sustainable Development Goal 3, Good Health and Well-Being, which includes the reduction of childhood mortality (Ndeezi *et al.,* 2016).

SCD is one of the most common inherited life-threatening disorders in humans; it predominantly affects people of African, Indian, and Arab ancestry. It is estimated that over 80% of over 300,000 annual births occur in sub-Saharan Africa (SSA), with the largest burden from Nigeria and the Democratic Republic of Congo. The gene frequency is highest in West African countries, with 1 in 4 to 3 (25–30%) being carriers of HbS, compared to 1/400 African Americans, and is variable in European populations. The prevalence of SCD in developed countries is increasing partly due to migration from high-prevalence countries (Inusa *et al.,* 2019). Particularly in Nigeria, a study was conducted to estimate the prevalence of SCD among children in Plateau State Specialist Hospital (PSSH), Jos, Nigeria. Results showed that the prevalence of SCD in PSSH, Jos, from 2012 to 2014 was 26.9/1000 population of pediatric patients. There was a gradual increase in the prevalence rate from 25.8/1000 in 2012 to 26.8/1000 in 2013 and 28.1/1000 in 2014 (Nanbur et al., Prevalence of Sickle Cell Disease among Children Attending Plateau Specialist Hospital, Jos, Nigeria, 2018)

A study in Tanzania revealed 9/50 (18%) participants were found to be sickling test positive, 5 (55.6%) of whom were male and 4 (44.4%) were female. Almost half of the SCD-positive children were between 25 and 36 months old. The factors that were strongly associated with death among these children included low haemoglobin levels and high total and conjugated bilirubin levels, with the highest incidence of death being reported among children under 10 years (Musyoka *et al.,* 2018). In Uganda, the gene prevalence of HbSS is 13%, with an estimated 20,000 children born with SCA annually. It is estimated that <50% of Ugandan children with SCA survive to their fifth birthday, whereas children with SCA in developed countries are expected to live up to 60 years. Delayed diagnosis, difficulties in accessing adequate care, and a shortage of trained health care professionals contribute to the high rate of early mortality (Namazzi *et al.,* 2017). This study aims to determine the Prevalence of sickle cell disease and sickle cell trait among children below 17 years of age attending Entebbe Regional Hospital, Wakiso district, Uganda.

Methodology.

Study design.

A cross-sectional and prospective study design was adopted and used to collect data from the respondents who attended the children's ward of Entebbe Regional Referral Hospital.

Study area.

The study was conducted at Entebbe Regional Referral Hospital. The hospital is located in the central business district of the town of Entebbe, in Wakiso District, approximately 44 kilometers (27 miles) by road, southwest of Mulago National Referral Hospital, the largest hospital in the country, located in Kampala, Uganda's capital and largest city. The coordinates of the hospital are: 0°03'50.0"N, 32°28'18.0" E (Latitude: 0.063874; Longitude:32.471655). The hospital provides a wide range of medical services, including pediatrics, radiology, laboratory, maternity, immunization, general surgery, internal medicine, orthopedics, and operating rooms.

Study population.

The target populations were children below the age of 17 years who attended Entebbe Regional Referral Hospital at the time of the study. The accessible populations were children below the age of 17 years who attended Entebbe Regional Referral Hospital at the time of the study and assented to the study.

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Selection Criteria.

Inclusion criteria.

The study only included children below the age of 17 who attended Entebbe Regional Referral Hospital at the time of the study and assented to the study.

Exclusion criteria.

Critically ill patients were excluded from the study.

Sample size determination

The sample size was determined using the formula, Kish and Leslie 1965; $N = PQZ^2/E^2$,

P = population estimate is unknown at this study site. Therefore, 9.7% (0.097) was taken (Hernandez *et al.*, 2021).

Q = 1-P (1-9.7)

Z = confidence level, 95% (1.96) was taken.

E = error level (precision), 10% (0.05). Was taken $N = \frac{0.097*(1-0.097) * 1.96^2}{0.05^2}$

$=134$

Therefore, 134 study respondents were included in this study.

Sampling technique.

A non-probability convenience sampling technique was used to recruit participants. Any child below the age of 17 years who attended Entebbe Regional Referral Hospital during the study period was selected for the study, regardless of gender. This technique is the choice because it is quick, easy to implement, and cost-effective.

Sampling procedure

Children below the age of 17 who attended Entebbe Regional Referral Hospital were approached. The objective of the study was explained to the parents of the children, and they were requested to allow their children to participate in the study. Various patients were approached until a minimum sample size of 97 was attained. Those who were considered eligible were requested to sign an informed consent form. Interview data were collected using an interviewer-administered questionnaire to obtain socio-demographic information. Thereafter, blood samples were collected from the participants for screening for sickle cell disease following the standard operating procedures.

Data collection methods.

Data was collected prospectively from the study participants by answering the researcher-administered questionnaire that included socio-demographic information, awareness, and knowledge of parents of the children about sickle cell disease and trait. Additionally, blood samples were obtained from the study participants, prepared, and then finally analyzed for sickle cell screening.

Data collection tools

Questionnaires were used to collect data on socio-demographic characteristics, awareness, and knowledge of parents of the children about sickle cell disease and trait. A simple data entry sheet was used to record laboratory test results for sickle cell solubility test results.

Data collection procedure.

The socio-demographic characteristics, awareness, and knowledge of parents of the children about sickle cell disease and trait were collected by using a semi-structured questionnaire prepared for this study. Data for these characteristics were collected by trained medical laboratory research assistants through a face-to-face interview. The study participants were interviewed after written informed consent was taken. After receiving informed consent from the study participants, their blood samples were collected, that was used to determine each participant's sickle cell status.

Solubility Test for Sickle Cell.

The solubility test is a rapid screening method used to detect the presence of hemoglobin S (HbS), the abnormal hemoglobin responsible for sickle cell disease.

Principle.

The sickle cell solubility test is based on the reduced solubility of deoxygenated hemoglobin S in a high-molarity phosphate buffer containing a reducing agent (e.g., sodium dithionite). Normal hemoglobin (HbA) remains soluble in the buffer. HbS, when deoxygenated by the reducing agent, precipitates out of solution due to its decreased solubility. This causes turbidity or cloudiness in the solution, indicating the presence of HbS. If the sample remains clear, it is negative (no HbS). If it becomes turbid, it is positive for HbS (either trait or disease).

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Reagents

1. Sodium dithionite (reducing agent)
2. Phosphate buffer (high molarity)
3. Saponin or detergent (for lysing red blood cells)
4. Test sample: Whole blood (preferably EDTA anticoagulated)

Procedure.

1. Label a clean test tube or test well.
2. Add 2 mL of solubility buffer solution
3. Add 20 µL of whole blood (EDTA)
4. Mix thoroughly to lyse the red cells and reduce the hemoglobin.
5. Let the mixture stand for 3–5 minutes at room temperature.
6. Observe the mixture:
   - Place a white background (e.g., paper) behind the test tube.
   - Look through the mixture from the side (not from the top).

Table 1 Interpretation.

Appearance	Result	Interpretation
Clear solution	Negative	No HbS or HbAS present (normal hemoglobin only)
Red precipitate	Positive	HbS present (SCD)
Pink precipitate	Positive	HbAS (SCT)

Study variables.

Independent variable

The independent variable was prevalence, knowledge, and awareness of sickle cell disease.

Dependent variable.

The dependent variable was sickle cell disease.

Quality control.

After the completion of each questionnaire, cross-checking was done between the data collector and principal investigator to ensure that all the data was collected. The label on the test tube and the study participants' unique identification number on the questionnaire were checked. Before patient sample processing, quality controls were performed, and the study participants' result was taken after confirmation that the control results were within an acceptable range.

Data analysis and presentation.

Data from the predesigned form and laboratory results were entered in books and then entered in a Microsoft Excel data sheet. Data analysis was done using SPSS version 20 with the help of a Biostastician. The analyzed data were presented as tables and figures with narratives under each. All statistical analysis of data was done at a 95% level of confidence and a level of error as 10%, error beyond this were considered statistically significant.

Ethical consideration.

Ethical approval for the study was obtained from the Mildmay Institute of Health Sciences Research Committee.

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An introductory letter from the Principal Tutor of Mildmay Institute of Health Sciences was obtained and presented at Entebbe Regional Referral Hospital. Approval to conduct the study was obtained at the study site. Informed consent was obtained from all recruited patients after a detailed explanation about the study in the language they understood best. Participation was voluntary, and each participant was given a consent form to fill out and sign. Confidentiality was maintained by using only patient numbers instead of names. The data generated was kept under lock and key, and soft copies were protected with a strong password. No unauthorized person was allowed to access the research data unless linked to the study. Participants' information and results were handled with confidentiality. If the findings were to be published, the identity of the participants would remain anonymous.

Results

Socio-demographic characteristics of the study participants.

Table 2: A table showing the socio-demographic characteristics of respondents

Variables	Category	Frequency (n=134)	Percentage (%)
Gender	Male	60	45.0
	Female	74	55.0
Total		**134**	**100**
Age (years)	**0-5**	55	41.0
	6-11	60	45.0
	12-17	19	14.0
Total		**134**	**100**
School level	Nursery	36	27.0
	Primary	67	50.0
	Secondary	12	9.0

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	Not schooling	19	14.0
Total		**134**	**100**

Table 2 shows the socio-demographic characteristics of the study participants. Findings showed that out of the 134 respondents, 55.0% (74/134) were females and 45.0 % (60/134) were males. The age range of respondents was 0–17 years. The majority of respondents, 45.0% (65/134), were aged 6-11 years, and most of the participants were attending primary school, 50% (67/134).

The prevalence of sickle cell disease among the study participants.

Table *3*: Showing the prevalence of sickle cell disease among the study participants.

Variables		**Category**	**Frequency**	**Percentage (%)**
Sickle cell Solubility test	**Positive**	**HbSS (SCD)**	8	6
		HbAS (SCT)	17	12.7
	Negative	**HbAA (Normal)**	109	81.3
	Total		**134**	**100**

Table 3 shows that the homozygous form, HbSS, was found in 8 respondents, giving a 6% (8/134) prevalence of sickle cell disease. Whereas 81.3% (109/134) of the respondents tested negative for both SCD and SCT

The Prevalence of sickle cell Trait among children below 17 years of age attending Entebbe Regional Hospital, Wakiso district, Uganda.

***Table* 4: Showing the Prevalence of sickle cell Trait among study respondents**

Variables		**Category**	**Frequency**	**Percentage (%)**
Sickle cell Solubility test	**Positive**	**HbSS (SCT)**	17	12.7

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		HbAS (SCD)	8	6
	Negative	**HbAA (Normal)**	109	81.3
	Total		**134**	**100**

Table 4 shows the heterozygous form; 17 study respondents were found to have the heterozygous form HbAS, giving a 12.7% prevalence of sickle cell trait. Whereas 81.3% (109/134) of the respondents tested negative for both SCD and SCT.

Findings showed that the Prevalence of sickle cell disease and sickle cell trait among children below 17 years of age attending Entebbe Regional Hospital was 18.7% (25/134).

Knowledge and awareness about sickle cell disease among parents of children below 17 attending Entebbe Regional Referral Hospital.

***Table 5*: Showing the knowledge of parents of respondents about sickle cell disease.**

Variable	**Response**	**Frequency (N=134)**	**Percentage (%)**
Heard about SCA/SCD before	Yes	**115**	**86.0**
	No	**19**	**14.0**
Has the child ever tested for SCD	Yes	**11**	**8.0**
	No	**123**	**92.0**
Know how a person can get SCD	Yes	**52**	**39.0**
	No	**82**	**61.0**
Sickle cell transmission	From parent	**87**	**65.0**
	I don't know	**23**	**17.0**
	Witchcraft/Cursed	**4**	**3.0**
	Others	**20**	**15.0**

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Know sickle cell	Yes	**103**	**77.0**
Crisis	No	**31**	**23.0**
Signs and symptoms of sickle	Pain	**97**	**72.0**
	Swelling of hands	**25**	**19.0**
	Others	**12**	**9.0**

Table 5 shows the knowledge and awareness of parents of respondents about sickle cell disease and sickle cell trait. One parent or guardian answered the researcher-administered questionnaire for each respondent. Findings showed that 86% (115/134) of respondents had heard about sickle cell. The majority, 92% (123/134), had children who had never been screened for sickle cell. Only 8% (11/134) of the children had ever been screened. Furthermore, 39% (52/134) of parents reported knowing the cause of sickle cell disease. The majority, 61% (82/134), were unaware of what causes SCD. A total of 65% (87/134) of parents reported that sickle cell is transmitted from parent to child. Seventeen percent (23/134) did not know how SCD is transmitted. Three percent (4/134) associated SCD with witchcraft, while 15% (20/134) gave other responses regarding transmission. The majority, 77% (103/134), knew about sickle cell crisis. Seventy-two percent (97/134) reported pain as a symptom of sickle cell, 19% mentioned swelling of hands and feet, and 9% (12/134) listed other symptoms.

Discussions.

Prevalence of sickle cell disease among study participants.

The study findings revealed an 18.7% (25/134) prevalence for both sickle cell disease and the prevalence of sickle cell disease SCD was 6% (8/134). The high prevalence was probably attributed to the low level of awareness and knowledge about sickle cell disease. Some of the respondents have financial constraints and cannot afford to pay for early sickle cell screening services, especially at the private health facilities. This finding agreed with a cross-sectional study conducted to determine the prevalence of sickle cell disease among children aged1-15 years at Luwero Hospital in the Luwero district. The results revealed that of the 96 children who participated in the study, 11 tested positive for SCD, giving a prevalence of 11.5% (Matovu, 2022)

Prevalence of sickle cell trait among study participants.

The study findings revealed an 18.7% (25/134) prevalence for both sickle cell disease and sickle cell trait, and the prevalence of sickle cell Trait (SCT) is 12.7% (17/134). The high prevalence was probably attributed to the low level of awareness and knowledge about sickle cell disease. Some of the respondents have financial constraints and cannot afford to pay for early sickle cell screening services, especially at the private health facilities. This finding agreed with the HbAS prevalence of 18.7% (42/225) obtained in a study carried out in Bungoma County, Kenya, to determine the prevalence of HbAS among adolescents (E.K Watenga *et al.,* 2020).

However, a prior study carried out to determine the burden of SCT and factors influencing the uptake of screening services among secondary school students in Uganda discovered a low prevalence of sickle cell trait, 5.8% had the HbAS genotype (carriers), and 0.9% for HbSS genotype (sicklers), which contrasts with our findings (Namukasa *et al.,* 2024).

The knowledge of parents of respondents about sickle cell disease.

Only 8% (11/134) of participants in our study reported having been previously tested for sickle cells. This might be due to the fact that screening for diseases is not something that people often consider doing, especially if they are not feeling ill. It could also be explained by the fact that SCD testing is only offered in a few specific medical facilities and is not a free service. In this study, most parents did not know the Hb genotype for their children. Similarly, a cross-sectional study among adults (mean age 29 years) in Muscat, Oman, showed that 36% thought SCT screening was a

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difficult and painful process, which explained the small percentage who had been screened (24.4%), and 37.8% felt it would be hard to convince their partners to go for screening (Al-Azri *et al*., 2016).

In another study, most (94.6%) youths (aged 22–29 years) in South Nigeria knew their Hb genotype (SCD carrier status), with the most common reason for checking their sickle cell status being a school entry requirement (Adewoyin AS *et al.,* 2015). Most participants in this study, 86% (115/134), had heard about sickle cell disease. This is presumably because parents of these children had easy access to tools like the Internet, and some children were also taught about hereditary illnesses at school. This finding was consistent with previous studies (Ameade EPK *et al.,* 2015), which reported that most participants learned about SCD from the media and school. This suggests that schools and the media can be effective institutions and platforms for educating people about SCD. Carriers of the disease who do not know their genotype may not seek the necessary genetic counseling to make informed marital choices. Therefore, school authorities should encourage parents to find out their children's Hb genotype, as well as students older than 18 years.

The majority of the parents of respondents, 65%, were aware that parents pass sickle cell to their children, but 23/134, 17%, were unaware of how SCD is transmitted. This may be because some parents may not know their Sickle cell status and have never had anyone with sickle cell disease in their family. In addition, a previous study reported that most Ugandans only discovered they had the sickle cell allele when they had a child with SCD, yet sickle cell is known to play a causal role in several morbidities, which makes the disclosure process critical (Harrison S.E. et al., 2017).

Similar findings were reported in another study conducted to determine the knowledge, perceptions, and practices towards sickle cell disease, in which more than half (54.0%) of the participants knew SCD was inherited from both parents, and the majority (62.9%) had a family member with SCD (Tusuubira SK *et al*., 2018).

Conclusion.

The study findings indicated that sickle cell disease and sickle cell trait remain important health concerns among children below 17 years attending Entebbe Regional Referral Hospital. This highlights the continued need for early identification, routine screening, and strengthened preventive strategies within the pediatric population.

Although many parents had heard about sickle cell disease, there were notable gaps in knowledge regarding its causes, inheritance patterns, and prevention. A large proportion of children had never undergone screening, suggesting missed opportunities for early diagnosis and appropriate management. While some parents were aware of common symptoms such as pain crises, misconceptions, and limited understanding about the condition were still evident among a number of respondents.

Recommendations.

There is a need to scale up screening services for sickle cell disease in the community so as to enable early diagnosis and treatment of the disease and reduce morbidity and mortality from it. Furthermore, the government, through the Ministry of health should set aside funds to facilitate awareness projects encouraging premarital testing of couples for sickle cell trait and make it mandatory to curb the increasing prevalence of SCD and provide enough funds for a large stock of medication for the Sicklers' to prevent early mortality and for better management and supervision.

Acknowledgment.

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List of abbreviations and acronyms

DNA: Deoxyribonucleic acid
G/dl: Grams per deciliter
Hb: Haemoglobin
HbAS/SCT: Sickle cell trait
HbSS/SCA: Sickle Cell Anemia
SCD: Sickle Cell Disease.
SPSS: Statistical Package for the Social Sciences.

Source of funding.

The study was not funded.

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Conflict of interest.

There is no conflict of interest.

Availability of data.

Data used in this study are available upon request from the corresponding author.

Authors contribution.

LN designed the study, conducted data collection, cleaned and analyzed data, and drafted the manuscript.
AS supervised all stages of the study from the conceptualization of the topic to manuscript writing and submission.
FA supervised the research process.
HN supervised the research process.
JFN supervised the research process.

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