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RESEARCH ARTICLE

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Morphological and Growth Profile of Children Suffering from Sickle Cell Disease in the Democratic Republic of the Congo: A Comparative Study of Statural, Weight, and Craniofacial Characteristics in Lubumbashi and Kinshasa

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ABSTRACT

Background: Sickle cell disease (SCD) remains one of the most prevalent genetic disorders in Sub-Saharan Africa. Beyond the well-documented hematological and infectious complications, the concept of a “sickle cell facies” has been long discussed by clinicians. However, this clinical impression has rarely been substantiated by scientific studies, particularly in the Congolese context.

Aim: This research aimed to determine whether measurable differences in growth and facial morphology could distinguish homozygous sickle cell children (HbSS) from their healthy peers (AA).

Methodology: We conducted a case-control study in Lubumbashi and Kinshasa, involving 85 children with confirmed homozygous sickle cell disease (HbSS) and 425 healthy AA controls, aged 6–13 years. Data collected included height, weight, head circumference, and various craniofacial measurements (nose, face, ears). Measurements were taken using standardized photo-anthropometry and analyzed statistically by comparing means, with a significance threshold set at $p<0.05$.

Results: Children with sickle cell disease presented with growth retardation from an early age. Their head circumference was smaller than that of the controls until age 8, after which this difference diminished. Morphologically, they had a wider and higher nose, with a significantly increased nasal index. Their faces were generally taller and wider, and their ears were also larger in height and width compared to the healthy children.

Conclusion: This study highlights the existence of a distinct morphological profile in Congolese children with sickle cell disease, confirming the clinical intuition of a “sickle cell facies.” These characteristics could, in the long term, enrich diagnostic tools and guide screening in settings where access to biological exams remains limited.

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Introduction

Sickle cell disease (SCD) is a hereditary hemoglobinopathy caused by a point mutation in the beta-globin gene, leading to the substitution of glutamic acid by valine at position 6 and the production of hemoglobin S (HbS) [1]. When this hemoglobin is deoxygenated, it polymerizes, causing the red blood cells to become rigid and fragile [2]. This cellular deformation leads to chronic hemolytic anemia, recurrent microvascular occlusion, and a wide range of acute and chronic complications [3].

With over 300,000 new cases each year, sickle cell disease is the most common monogenic disorder worldwide [4]. Sub-Saharan Africa accounts for nearly 75% of sickle cell births [5]. According to the World Health Organization (WHO), 2% of newborns in Central Africa and up to 25% in some regions of West Africa carry the mutation [6]. In the Democratic Republic of Congo (DRC), approximately 25% of the population is heterozygous (AS), and the annual incidence of homozygous forms (SS) ranges between 15 and 25 per 1,000 live births [7].

Despite progress in medical care, the mortality rate for sickle cell disease remains high in low-resource countries. Before the age of five, more than 50% of affected children die from infectious, anemic, or vaso-occlusive complications due to a lack of early screening and adequate care [8,9]. However, in countries with neonatal screening programs and specialized follow-up, patients' life expectancy can exceed 50 years [10].

In addition to its hematological and infectious manifestations, sickle cell disease is associated with morphological and skeletal abnormalities. Recurrent medullary crises, growth disorders, and chronic hypoxia can impair both linear growth

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and craniofacial morphology [11,12]. Clinicians have long reported the existence of a "sickle cell facies" characterized by a broadened face, a flattened nose, and sometimes dentofacial anomalies [13]. However, these observations have often been based on subjective clinical impressions rather than quantitative data [14].

Some anthropometric studies have shown that children with sickle cell disease exhibit significant statural and weight retardation as well as craniofacial variations, particularly in the nose and head circumference [15,16]. However, most of these studies have been conducted in West Africa. There is few data available from Central Africa, particularly the DRC [17]. The prevalence of the Bantu haplotype, associated with more severe disease, could confer unique morphological characteristics to Congolese patients [18].

Thus, a better understanding of the anthropometric characteristics of sickle cell children could serve a dual purpose: enriching knowledge about the phenotypic expression of the disease and providing complementary clinical screening tools in contexts where electrophoresis and molecular biology are not always accessible [19,20].

Methodology

This study was designed as a multicenter case-control survey conducted between December 2015 and May 2016 in two cities of the Democratic Republic of Congo (DRC): Lubumbashi and Kinshasa. Sickle cell patients were recruited at the Sickle Cell Care Center (CPCD) of Sendwe Hospital in Lubumbashi and the Sickle Cell Anemia Center (CEMAS) in Kinshasa, both referral centers specializing in the management of hemoglobinopathies. The control group consisted of healthy children recruited from public and private primary schools in Lubumbashi.

Study Population

The cases were children aged 6 to 13 years diagnosed with homozygous sickle cell disease (HbSS) confirmed by hemoglobin electrophoresis. The controls were children of the same age group with the HbAA genotype, recruited based on strict criteria: absence of chronic diseases, malformations, or obvious facial dysmorphisms, and Congolese Bantu ancestry for at least two generations to limit the influence of ethnic variability. Informed consent was obtained from the parents for each participant. For the controls, a note was placed in the children's class journals, and only those for whom parental consent and child assent were obtained were included in the study.

Study Parameters

The data collected included: height (measured with a calibrated stadiometer), weight (measured with an electronic scale), head circumference (measured with a tape), and various craniofacial measurements obtained through photo-anthropometry. Photographs were taken from both frontal and lateral views on a grid background for scaling. The cephalometric landmarks included: facial width (zy-zy), facial height (tr-gn), nasal width (al-al), nasal height (n-sn), nasal index (al-al/n-sn × 100), and ear height (sa-sba).

Data Collection

Data were collected using a standardized, pre-tested data collection form, which included several key variables: sociodemographic, clinical, anthropometric, and morphological data.

- Sociodemographic variables included age, sex, and the presence of a caregiver.
- Clinical variables included the sickle cell disease diagnosis method (HbSS confirmed by hemoglobin electrophoresis), a history of vaso-occlusive crises and other complications, prophylactic treatments (such as cotrimoxazole and penicillin), and current treatment regimens, including the use of hydroxyurea.
- Anthropometric measurements included height, weight, and head circumference, all measured with calibrated equipment. Craniofacial measurements were obtained using a standardized photo-anthropometry protocol, with frontal and lateral photographs taken. The specific facial and nasal measurements included facial width (zy-zy), facial height (tr-gn), nasal width (al-al), nasal height (n-sn), and nasal index (al-al/n-sn × 100). Ear height was also measured, along with other morphological parameters.

All photographs were taken on a grid background to ensure a uniform scale for precise measurements. Each measurement was repeated three times to ensure consistency and accuracy. Photogrammetry, used in other studies on craniofacial morphology, offers the advantages of being simple, reproducible, and low-cost, even though it remains limited by the sometimes approximate identification of certain landmarks [21,22].

Data Processing and Statistical Analysis

The collected data were entered into Microsoft Excel 2016, cleaned for any inconsistencies, and then analyzed using SPSS version 25.0. Descriptive statistics were used for continuous variables (such as height, weight, and head circumference), with means and standard deviations reported. Categorical variables were analyzed using frequency distributions. For bivariate analysis, comparisons between the two groups (HbSS and HbAA) were performed using the Student's t-test for continuous variables and the Chi-square test for categorical variables. Finally, multivariate analysis was conducted to explore factors associated with craniofacial morphology and growth, adjusting for variables such as age, sex, treatment, and the presence of complications.

Ethical Considerations

The study was approved by the Ethics Committee of the Faculty of Medicine at the University of Lubumbashi. Informed consent was obtained from the parents or legal guardians of each participant, and children's assent was also obtained where appropriate. For the control participants, an information note was sent through the children's school journals, and only those for whom we received both parental consent and the child's assent were included in the study. Confidentiality was strictly maintained throughout the process, and all data were anonymized before analysis.

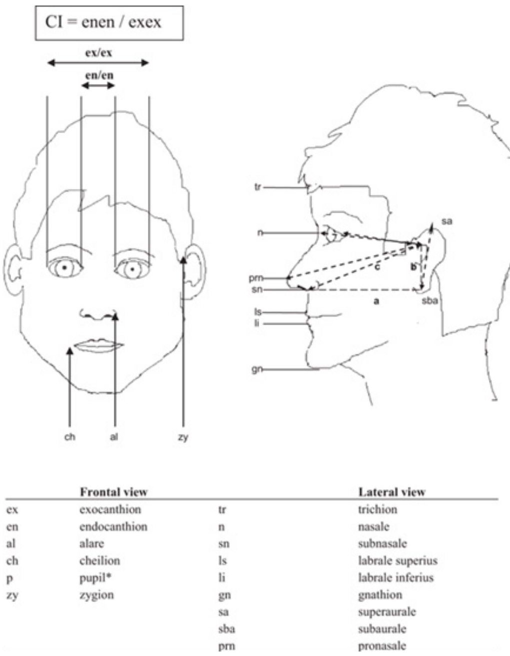


Figure 1: Anthropometric landmarks of the face in frontal and lateral views [26].

Results

Sociodemographic Characteristics

A total of 85 children with homozygous sickle cell disease (HbSS) were enrolled in the study, along with 425 healthy children (HbAA) from the same age group (6-13 years). The mean age of the sickle cell group was 9.7 years (± 2.4), with 54% male participants. Among the controls, the mean age was 9.5 years (± 2.2), with 52% male participants. The gender distribution was comparable between the two groups, with no significant differences in age (p = 0.55).

Anthropometric Data

Children with sickle cell disease exhibited significantly lower height and weight compared to the control group. The mean height in the sickle cell group was 123.4 cm (± 12.3) compared to 130.2 cm (± 10.5) in the control group (p < 0.01). Similarly, the mean weight for sickle cell children was 25.3 kg (± 6.9), while it was 30.1 kg (± 7.4) in the control group (p < 0.01). These results reflect the overall growth retardation observed in the sickle cell cohort, consistent with known clinical manifestations of SCD. This information is summarized in Table 1.

Head Circumference

The average head circumference for sickle cell children was significantly smaller than that of controls. The mean head circumference was 51.4 cm (± 2.8) in the sickle cell group, compared to 53.6 cm (± 3.2) in the control group (p = 0.02). The difference in head circumference was most notable in children aged 6-8 years, with a trend toward normalization in older children (9-13 years), suggesting some potential recovery in cranial growth as the children aged. These observations are also included in Table 1.

Craniofacial Measurements

The craniofacial measurements showed significant differences between the two groups. Children with sickle cell disease had a wider and higher nose (mean nasal width = 3.9 cm, nasal height = 4.7 cm) compared to the controls (mean nasal width = 3.4 cm, nasal height = 4.3 cm) (p < 0.05). Additionally, the nasal index in the sickle cell group was higher (mean = 91.1) compared to the control group (mean = 85.4) (p < 0.01).

The facial dimensions were also altered. The facial height in sickle cell children was significantly increased, with a mean of 11.8 cm compared to 10.9 cm in the control group (p = 0.03). Similarly, facial width was also greater in the sickle cell group (mean = 14.5 cm) than in controls (mean = 13.2 cm) (p = 0.04). Table 1 shows these observations in detail.

Ear Dimensions

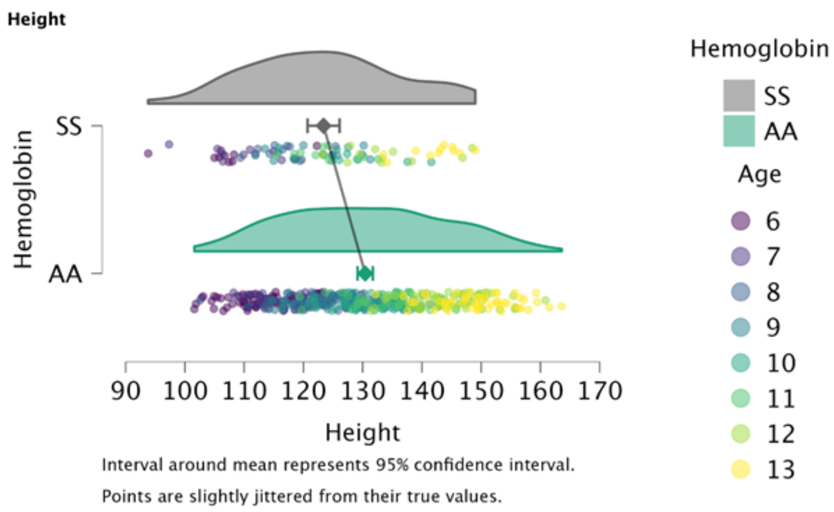
Ear measurements also differed between the two groups. The ear height of sickle cell children was notably larger (mean = 5.5 cm) than that of controls (mean = 5.1 cm) (p = 0.02). This variation may reflect compensatory mechanisms associated with the altered facial and cranial structures observed in sickle cell disease. These observations are also included in Table 1.

Growth Patterns and Puberty

Children with sickle cell disease exhibited growth retardation from an early age, with a significant catch-up in weight around the age of 10-11 years. However, this was followed by a subsequent deceleration in growth during the early stages of puberty, a phenomenon less commonly described in the literature. This trend suggests that while some degree of weight and height recovery occurs in pre-adolescence, the onset of puberty may be associated with a regression or stagnation in growth, likely due to the increased metabolic demands and

Table 1: Comparison of key measurements between SS cases and AA controls.

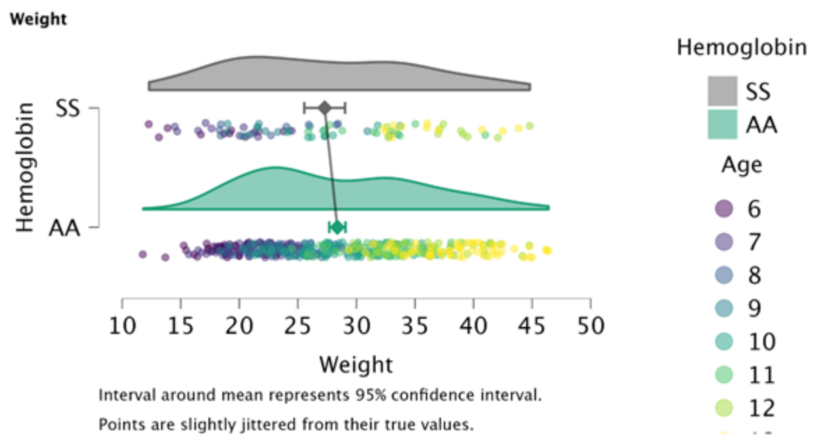
Parameters	SS (n=85), mean ± ET	AA (n=425), mean ± ET	Difference (SS–AA)	IC95 %	p-value
Height (cm)	134.2 ± 10.6	142.3 ± 9.8	−8.1	−9.7 à −6.5	<0.001
Weight (kg)	28.1 ± 6.2	31.3 ± 6.0	−3.2	−4.4 à −2.0	<0.001
Head circumference (cm)	50.1 ± 1.9	50.9 ± 1.8	−0.8	−1.2 à −0.4	0.001
Facial width, zy–zy (mm)	122.5 ± 5.4	117.3 ± 5.1	+5.2	+4.3 à +6.1	<0.001
Face height, tr–gn (mm)	173.6 ± 7.8	165.8 ± 7.5	+7.8	+6.5 à +9.1	<0.001
Nasal width, al–al (mm)	37.8 ± 3.4	34.6 ± 3.1	+3.2	+2.5 à +3.9	<0.001
Nose height, n–sn (mm)	47.2 ± 3.6	44.9 ± 3.5	+2.3	+1.4 à +3.2	0.002
Nasal index (al–al / n–sn ×100)	79.4 ± 6.8	74.2 ± 6.1	+5.2	+3.8 à +6.6	<0.001
Auricular height, sa–sba (mm)	59.3 ± 3.1	55.7 ± 2.8	+3.6	+2.9 à +4.3	<0.001
Auricular width (mm)	32.8 ± 2.9	30.2 ± 2.7	+2.6	+1.7 à +3.5	0.004



Heigth									
								95% CI for Mean Difference	
Primary Factor	*N*	Lower Whisker	25th Percentile	Median	75th Percentile	Upper Whisker	Mean	Lower	Upper
AA	425	101.60	119.80	129.80	140.00	163.60	130.43	129.1	131.7
SS	85	93.80	115.00	123.30	131.30	149.00	123.40	120.7	126.1

Note. N_{Total} = 510.
Interval around mean represents 95% confidence interval.

Figure 2: Comparative height growth chart.



Weigth									
								95% CI for Mean Difference	
Primary Factor	*N*	Lower Whisker	25th Percentile	Median	75th Percentile	Upper Whisker	Mean	Lower	Upper
AA	425	11.80	22.60	27.40	33.50	46.40	28.37	27.68	29.07
SS	85	12.30	20.80	27.20	33.20	44.80	27.29	25.55	29.03

Note. N_{Total} = 510.
Interval around mean represents 95% confidence interval.

Figure 3: Comparative weight growth chart.

hormonal changes during this period. These observations are illustrated by Figures 2, 3.

Discussion

Growth Retardation in Children with Sickle Cell Disease

This study, conducted in Lubumbashi and Kinshasa, highlights significant and reproducible differences between homozygous sickle cell children (SS) and their AA counterparts in terms of statural and weight growth, as well as craniofacial morphology. Studying children between 6 and 13 years old allows for the focus on a period of relatively homogeneous development, before major pubertal changes (accelerated growth, hormonal and morphological changes), thus minimizing the effects of puberty as a confounding factor on weight, height, and other anthropometric indicators.

Our results show that sickle cell patients have marked growth retardation, a wider and taller face, a widened nose with an increased nasal index, and larger ears. These findings confirm the existence of a distinct morphological phenotype, supporting the clinical hypothesis of the "sickle cell facies," a term that has been empirically described for several decades [13,15].

Comparison with Existing Literature

The growth anomalies observed in our cohort align with conclusions from studies conducted in West Africa and North America. Platt et al. had already demonstrated that children with sickle cell disease present with lower height and weight compared to their AA peers, with growth retardation starting in early childhood [23]. Similar results were reported in Nigeria, where Odetola described significant staturo-ponderal retardation in SS patients [24]. Our study confirms these findings in the Congolese context, but also highlights a specific profile: the weight catch-up observed around 10–11 years, followed by a pubertal deceleration, a phenomenon less commonly described elsewhere. This catch-up followed by pubertal deceleration could suggest a therapeutic window during which nutritional support and more targeted medical follow-up could maximize growth and improve the health of sickle cell children.

Craniofacial Findings

Few quantitative studies have previously documented the sickle cell facies. In Egypt, studies showed nasal widening and altered facial profiles in SS children [11]. In Central Africa, Kaimbo et al. studied some orbital measurements in Zairian children, but without distinguishing hemoglobin status [25]. Our study is one of the first to demonstrate, through standardized photo-anthropometry, the existence of significant differences in facial width and height, nasal index, and auricular dimensions. These results confirm, with objective data, clinical impressions that were previously based on the experience of practitioners [12,13].

Pathophysiological Explanations

Several pathophysiological mechanisms may explain these anomalies. Chronic hemolytic anemia and recurrent tissue hypoxia are known to affect linear and weight growth [2,3]. Medullary hyperplasia, a consequence of ineffective erythropoiesis, leads to modifications in craniofacial morphology, with expansion of bone structures, particularly in the nasal and maxillary areas [11,17]. Additionally, the predominance of the Bantu haplotype in the DRC, characterized by particularly low production of fetal hemoglobin, could amplify the severity of morphological alterations compared to patients with more protective haplotypes (such as Senegalese or Arab) [18,19].

Clinical Implications

The results of this study have several clinical and public health implications. First, recognizing the sickle cell facies could serve as a preliminary clinical tool for suspicion, particularly useful in rural areas where hemoglobin electrophoresis is not accessible. In such contexts, morphological examination, combined with family history, could help clinicians make a presumptive diagnosis and expedite management [8,9].

Second, the identification of specific craniofacial anomalies could provide new indicators for longitudinal patient follow-up. A high nasal index or disproportionate facial growth could become indirect markers of disease severity to be explored further. Finally, these data open the door to the use of artificial intelligence (AI) and facial recognition as innovative tools for early screening, an approach recently tested in other genetic diseases [27].

Strengths and Limitations

Our study has several strengths. It is one of the largest comparative cohorts in Central Africa, with a substantial sample of AA controls, allowing for a robust comparison. The standardized photo-anthropometry methodology ensures reproducibility and accessibility in low-resource settings [21,22]. Furthermore, the statistical analysis included adjustments for age and sex, as well as a multivariate approach, ensuring the validity of the results.

However, some limitations must be acknowledged. Photo-anthropometry does not capture all three-dimensional landmarks, unlike 3D cephalometry [22]. The study is cross-sectional, and therefore does not provide information on the long-term individual morphological evolution. Finally, the results are specific to a Congolese Bantu population and may not be generalizable to other ethnic or geographic contexts [6,20].

Perspectives

Our results call for the continuation of multicenter and longitudinal research. The integration of 3D cephalometry or digital imaging would allow for more accurate validation of our observations. Furthermore, the development of automated analysis software, using AI to detect facial morphological abnormalities, could revolutionize screening in rural areas of Sub-Saharan Africa. Finally, our data underline the need to strengthen neonatal screening programs and growth monitoring in children with sickle cell disease to reduce the morbidity and mortality associated with this condition [7,10].

Conclusion

This study provides clear and statistically robust evidence that Congolese children with sickle cell disease exhibit a distinct morphological profile. Our findings confirm the existence of an objectifiable sickle cell facies, which opens new possibilities for screening and monitoring. These results highlight the importance of incorporating morphological assessment as a complementary clinical tool in the early detection of sickle cell disease in Sub-Saharan Africa.

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