

Ocular manifestations of sickle cell disease in heterozygous subjects: Senegalese experience

Ka AM^{1*}, Ba EA², Wane AM², Sow AS², Diagne JP¹, De Medeiros ME¹, Kane H², Dieng M², Mouen Bengue G², Ndiaye JMM², Sow S¹, Nguer M², Ndoye Roth PA², Ndiaye PA¹

¹Department of Ophthalmology of Abass Ndao Hospital, Dakar (Senegal)

²Department of Ophthalmology of Aristide Le Dantec Hospital, Dakar (Senegal)

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*Corresponding author: Aly Mbara KA, Department of Ophthalmology of Abass Ndao Hospital, Dakar (Senegal), Phone: 00221775588815; Fax: 00221338420023; E-mail: kaam75@hotmail.com

Abstract

Introduction: Sickle cell disease is the most widespread hemoglobinopathy in the world and affects nearly 10% of the population in sub-Saharan Africa. It is responsible for ophthalmological manifestations very little described by the literature in our country. We report our experience of ocular lesions encountered in heterozygous sickle cell subjects monitored in our department.

Material and methods: From June 2004 to July 2011, we initiated a retrospective study which allowed us to recruit sickle cell patients for a research on ocular involvement. Each patient had complete ophthalmologic examination and fluorescein angiography. The descriptive analysis was carried out using the EPI-INFO software version 6.04.

Results: Out of 80 patients received, eight were retained with a sex ratio of 0.6%. The mean age was 34 years for extremes of 22 and 49 years. It was 50% AS sickle-cell anemia and 50% SC. On examination, the visual acuity (VA) varied from the absence of luminous perception to 10/10. The lesions found were bilateral in 7 cases and unilateral for 1 case.

Comments: Sickle cell disease is a hemoglobinopathy which predominates in the black subject and the male gender is the most affected. The SC genotypic form is the main provider of sickle cell retinopathy. Moreover, the signs are more marked as the subject is older, whereas this intensity varies inversely according to the genotype; it is more important in the SC form than in AS and finally SS form.

Conclusion: The ocular lesions related to sickle cell disease are serious and involve the functional prognosis of the eye especially of young and black subjects.

Keywords: Sickle Cell Disease; Eye; Black Race

Introduction

Sickle cell disease is a genetic disease with autosomal recessive transmission due to the presence in the red blood cell of abnormal hemoglobin called hemoglobin S. It would be the most widespread hemoglobinopathy in the world affecting nearly 120 million people according to the WHO. This disease is very frequent in Africa south of Sahara, where in Senegal it affects almost 10% of the population. It is responsible for polymorphic clinical manifestations, in particular ophthalmologic. However, the latter are very little described by the literature in our country. We report our experience of ocular lesions found in heterozygous sickle cell subjects monitored in our department.

Material and methods

From June 2004 to July 2011, we initiated a retrospective study of sickle cell patients for screening for ocular disorders

related to this disease. Each patient had a complete ophthalmologic examination (both socio-demographic and clinical) and fluorescein angiography. The sickle cell disease form was confirmed by the electrophoresis of hemoglobin. The descriptive analysis was carried out using the EPI-INFO software version 6.04.

Results

From June 2004 to July 2011, 80 patients were examined, and among them 8 were selected. Our sample consisted of 3 men and 5 women. The mean age was 34 years for extremes of 22 and 49 years. The genotypic forms were SC in 50% of cases and AS in 50% of cases.

On examination, the VA varied between the absence of light perception at 10/10. At the level of the anterior and posterior segments lesions were bilateral in 7 cases (all AS and in 3 SC cases) and unilateral for 1 SC case. (Tables 1 and 2).

Table 1: Characteristic of our study population

N°	Age	Gender	Form	VA		Lesions	
				RE	LE	RE	LE
1	49	F	SC	10/10	1/10	Normal	Peripheral lesions of vitreoretinopathy with vitreous traction on upper retinal vessels and peripheral ischemia
2	22	M	AS	PL+	CLD 1.5m	Traction flanges, peripheral ischemia, sea fan and total cataract	Ischemia and peripheral traction lesions, muscular dystrophy, retinal pan photocoagulation scars, and retinal vessels
3	28	F	AS	1/10	1/10	Liquefaction of vitreous, lumps of pigment in rosary with a hole at 7H on the equator in the lower nasal and choroiditis	Liquefaction of the vitreous
4	25	M	AS	PL+	1/10	Liquefaction of the vitreous, RD scarred at 7H, RD with tear at 4H-5H, hemorrhages at 1H and laser impacts at 9H-10H	Lower and temporal RD with VRP
5	27	M	AS	PL+	1/10	RD with demarcation line determining macular folds, pigment clods. Healthy retina between 11H and 12H	Pigment clods, pseudo phakia
6	38	F	SC	2/10	3/10	Pigment clods, vitreoretinal proliferation	Even lesions
7	43	F	SC	PL+	9/10	Sectoral iris atrophy posterior capsular obstructed cataract	Sectoral atrophy posterior capsular cataract, presence of upper temporal ischemic zones and neo-vessels
8	40	F	SC	10/10	PL-	Fine neo-vessels, pupillary seclusion	Fine neo-vessels and dehiscence of the retina

VA: Visual Acuity, **CF:** Counts the fingers, **RD:** Retinal detachment, **PL+:** Positive light perception, **PL-:** Negative light perception, **Form:** type of sickle cell disease, **VRP:** Vitreoretinal proliferation, **RE:** right eye, **LE:** left eye, **M:** male, **F:** female

Table 2: Frequency of damage to the posterior segment

Lesion Form	Vitreous Liquefaction	Ischemia, Neo vessels	Pigment blocks	VRP	Holes, tears	RD	Gender
AS	2 Cases (11.76%)	1 Case (5.88%)	2 Cases (11.76%)	1 Case (5.88%)	2 Cases (11.76%)	2 Cases (11.76%)	3 M, 1 F
SC	1 Case (5.88%)	3 Cases (17.64%)	1 Case (5.88%)	2 Cases (11.76%)	0 Case	0 Case	4 F

RD: Retinal detachment, **VRP:** Vitreoretinal proliferation, **M:** male, **F:** female

Comments

Sickle cell disease is a hemoglobinopathy which predominates in black subject [4,5]. According to various authors cited by Sanchez [5] such as Diallo and Wade, and also Pichard, Resnikoff and Gentillini, the overall prevalence varies from 5 to 20% in West Africa. Prevalence peaks of 40% have even been reported in Central Africa [5]. Moreover, the male gender would be more affected according to Fox PD [6], however in our sample there

were 62.5% of women. The average age of our patients was 34 years. Indeed, the ocular manifestations of sickle cell disease are observed especially in the young adult. Thus Traore J. et al [7] find predominance between 26 and 35 years. Similarly, Fox PD et al [8] and Downes SM [9] find themselves respectively 20-39 years and 24-26 years.

Furthermore, the predominance of AS and SC forms in our serie is similar to the findings of Sanchez [5] who report that 8.4% of the Senegalese population is AS, 17% AC and 1% SS.

On the symptomatologic level, we did not find the classical conjunctival signs (pale and jaundice). This is probably related to the absence of the SS form in our serie. In fact, these signs are characteristic of sickle cell anemia in this form [5]. Our patients with SC phenotype received in consultation had VRP in half of the cases and a retinal ischemia in 3 quarters of the cases. According to Yehouessi [10], 65.4% of cases of proliferative retinopathy are found in SC sickle-cell disease. In their series Balo and coll [11] found an overall prevalence of sickle cell retinopathy of 68%. Based on these results, it should be noted that the SC genotypic form is therefore the provider of retinopathy, especially in its proliferative form.

Otherwise, two (2) AS patients had retinal detachment (RD). This complication, which constitutes the ultimate stage, has a relative frequency of 1 to 4% [12]. In general, SC patients are most affected by retinal detachment. Traore found 25.92% of retinal detachment in SC patients and John found it in 10% of SC subjects [14]. Kabo [15] found a single case of retinal detachment in an AS patient. The presence of retinal detachment in our patients, especially carriers of the sickle cell trait, should be a wake-up call for practitioners. In fact, retinal damage in AS patients exists and are potentially serious.

However a study conducted in Canada in 2003 showed that healthy sickle cell trait carriers did not have an increased risk of retinopathy [16]. Morel also mentions the rarity of ocular involvement in AS patients [17].

Moreover, the signs would be more marked as the subject is older. Binaghi M and Levy C [18] and Fox PD and coll [6] also find this trend. On the contrary, this intensity varies inversely according to the genotype; It is more important in SC form than in AS and finally in SS [6, 18, 19, 20, 21, 22]. The SC form would be the largest provider of ocular manifestations followed by the SS form and finally the AS form [18, 20, 22]. However, Fany A and coll [23] found that ocular lesions of sickle cell disease were frequent in the AS form but less pronounced than in the SC and SS forms. In our serie, the signs of vitreoretinal proliferation and peripheral retinal ischemia were observed at the same degree as in SC patients than in AS patients.

Conclusion

The ocular lesions associated with sickle cell anemia have been the subject of numerous publications. They are severe and involve the functional prognosis of the eye especially of young and black subjects. Their insidious evolution is always a cause of irreversible blindness, especially in our regions where delays in diagnosis are deplored. However, ocular signs in sickle cell anemia are not uncommon or less severe.

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