CASE SERIES:AUTOIMMUNE POLYENDOCRINOPATHY SYNDROME TYPE 1

DR.ANISHA RATHOD

FELLOW -PEDIATRIC OPHTHALMOLOGY AND STRABISMUS

CO-AUTHORS - DR. ASHISH DOSHI, DR.PRACHI AGASHE

DEPARTMENT OF PEDIATRIC OPHTHALMOLOGY AND STRABISMUS

K.B.H. BACHOOALI EYE HOSPITAL , MUMBAI

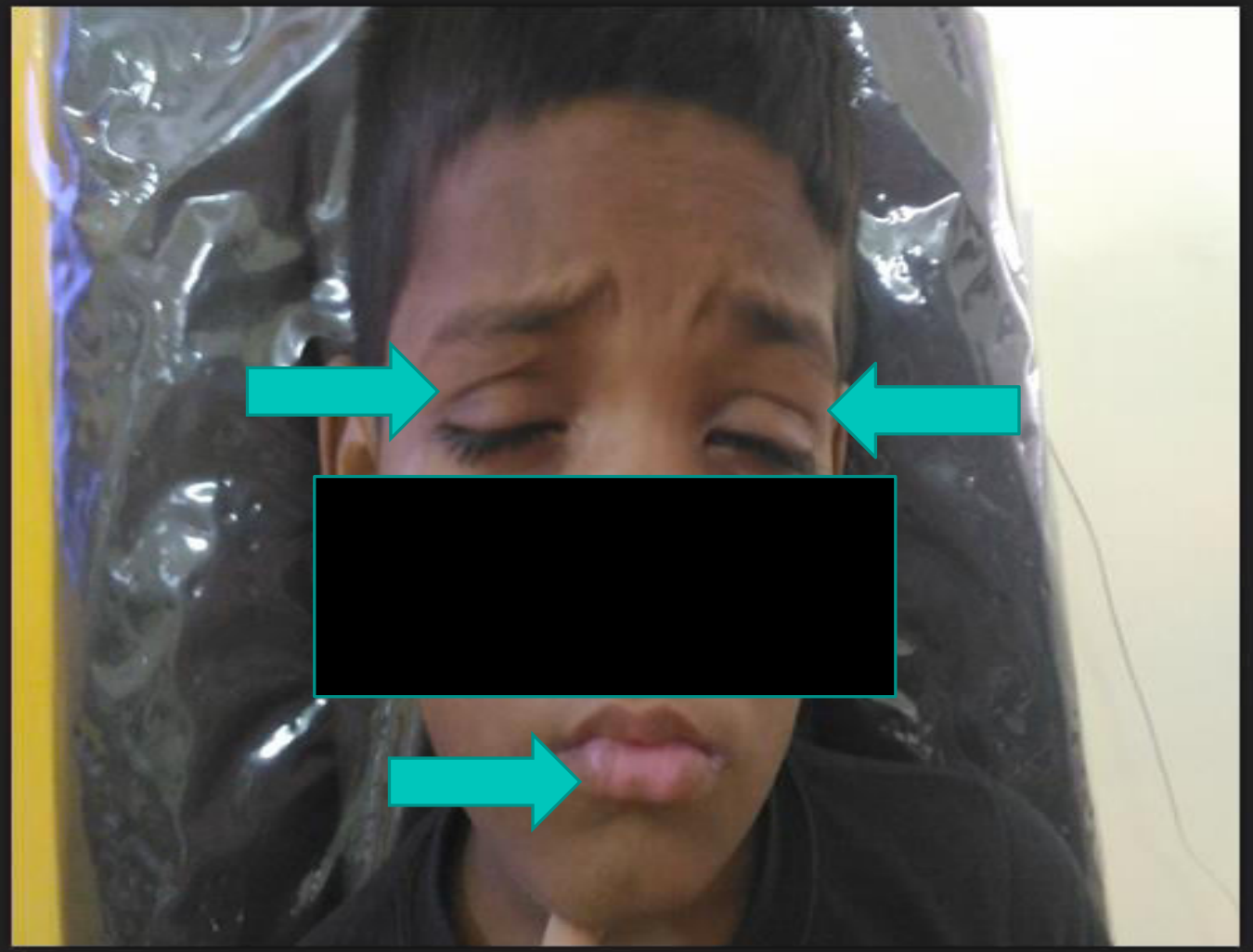
INTRODUCTION

- Rare autosomal recessive disorder that develops in early childhood.
- Hypocortisolism, hypoaldosteronism, delayed puberty, premature ovarian failure, hypoparathyroidism, hypocalcemia .
- APS-1 is clinically defined as the presence of at least two components of the classic triad of hypoparathyroidism, adrenal insufficiency, and mucocutaneous candidiasis

1.Eisenbarth GS, Gottlieb PA. Autoimmunepolyendocrine syndromes. N Engl J Med. 2004; 350:2068-79. 2. Perheentupa J. Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy. J Clin Endocrinol Metab. 2006; 91:2843-50. 3. Alimohammadi M, Björklund P, Hallgren A, et al. Autoimmune Polyendocrine Syndrome Type 1and NALP5, a Parathyroid Autoantigen.N Engl J Med. 2008; 358:1018-28.

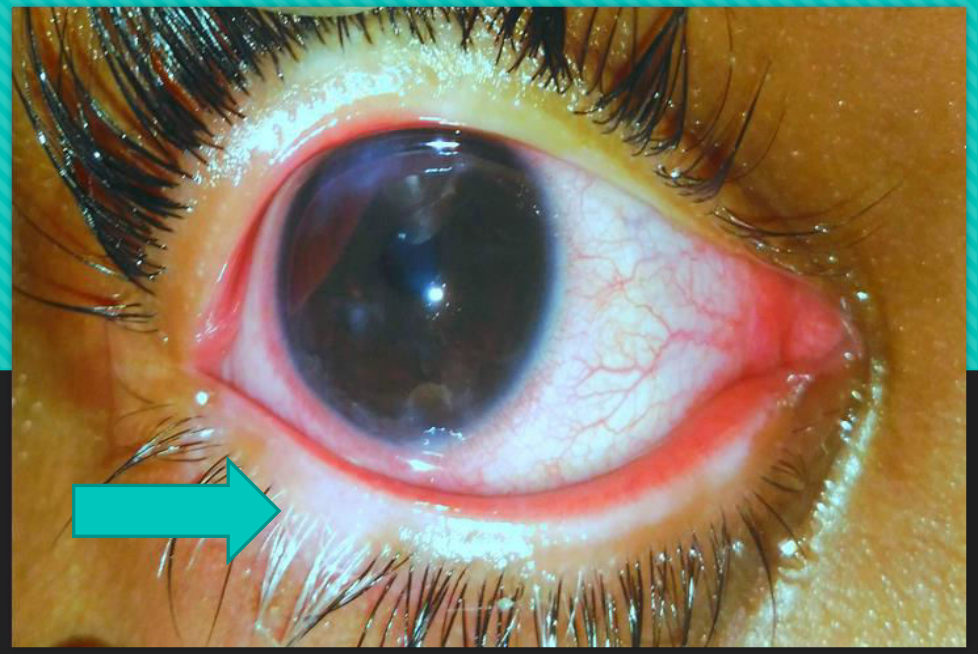
CASE REPORT

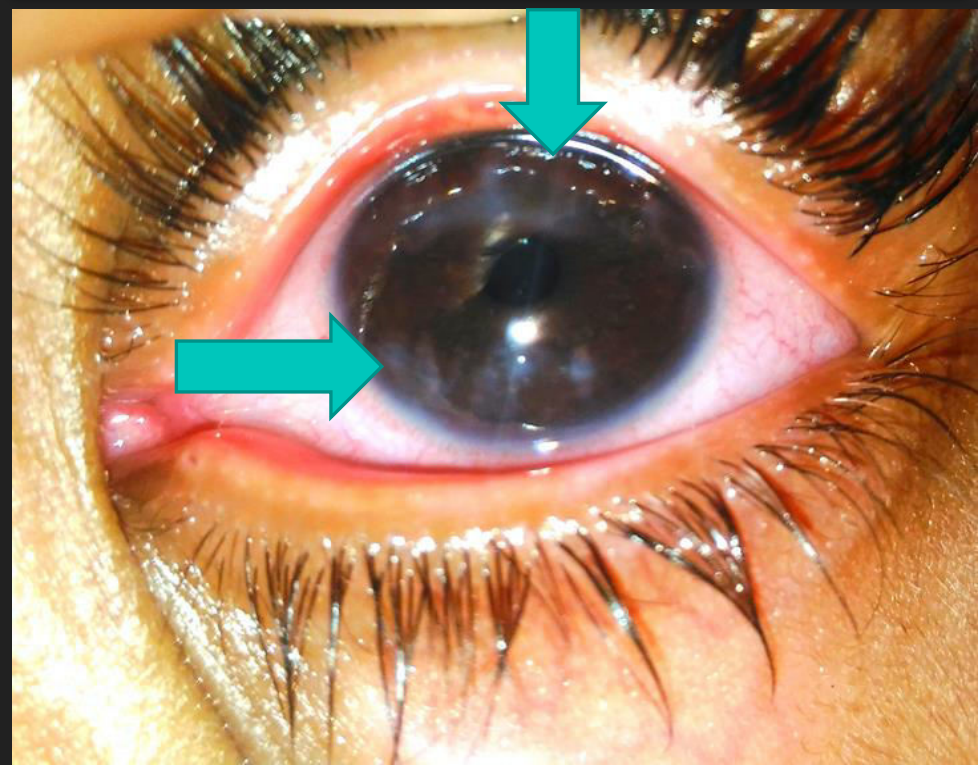
- A 8 year old male presented for examination with severe photophobia
- No History of associated redness, pain, discharge.
- He had similar episodes in the past .
- Episodes of hypocalcemic tetany.
- Oral candidiasis
- No History of ocular surgery or trauma.



OCULAR FINDINGS

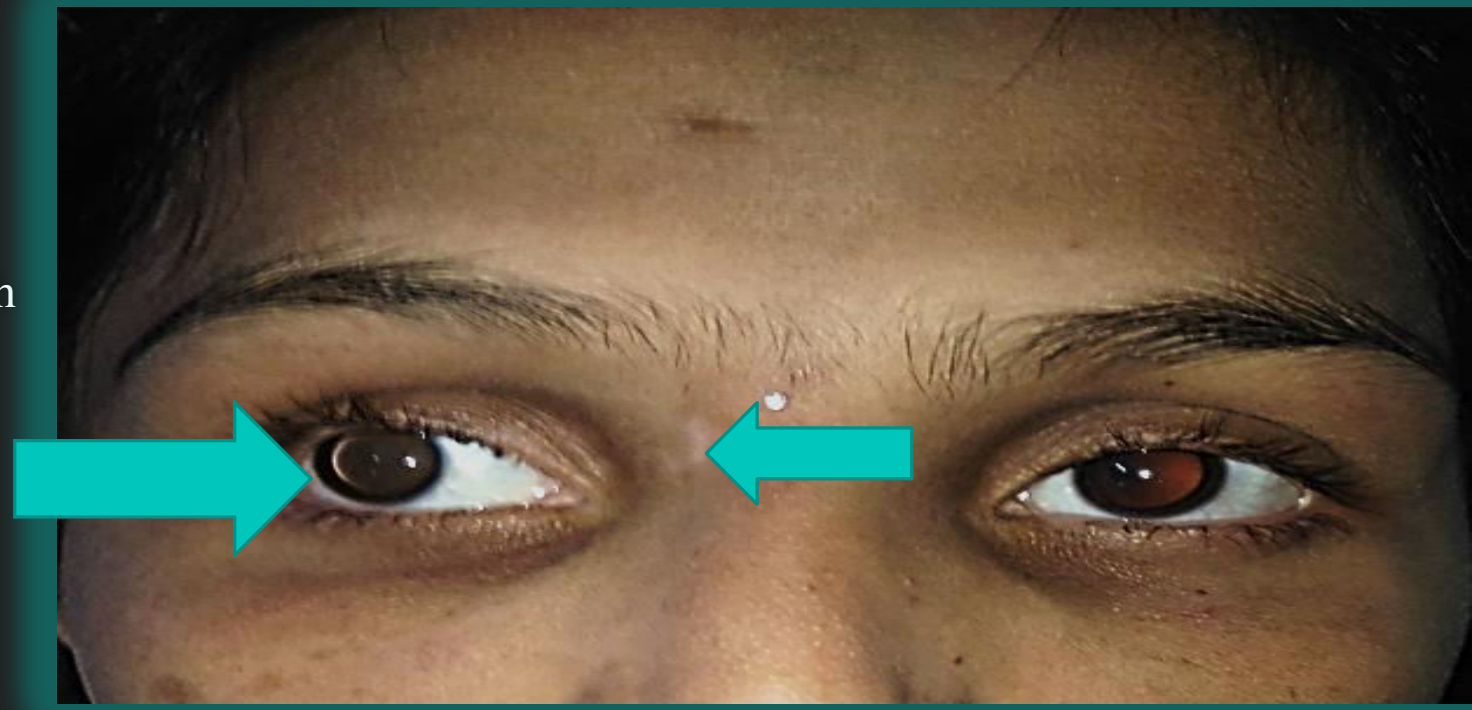
- Uncorrected visual acuity was 6/60 and 6/9 of right and left eye respectively.
- Anterior segment examination.
- Poliosis with vitiligo, mild conjunctival xerosis with corneal superficial lesions with superficial and deep vascularisation, Salzman nodular degeneration decreased corneal sensitivity,decreased tear film height.
- Schirmer test 5 mm and 10 mm of right and left eye respectively.
- Fundus-Normal
- No significant refractive error





CASE 2

- A 22 year old female presented for examination with progressive,painless dimunition of vision since 2 years
- No History of associated redness, pain, discharge.
- Adrenal insufficiency
- Hypoparathyroidism
- Mucocutaneous candidiasis
- Premature ovarian failure
- No History of ocular surgery or trauma.

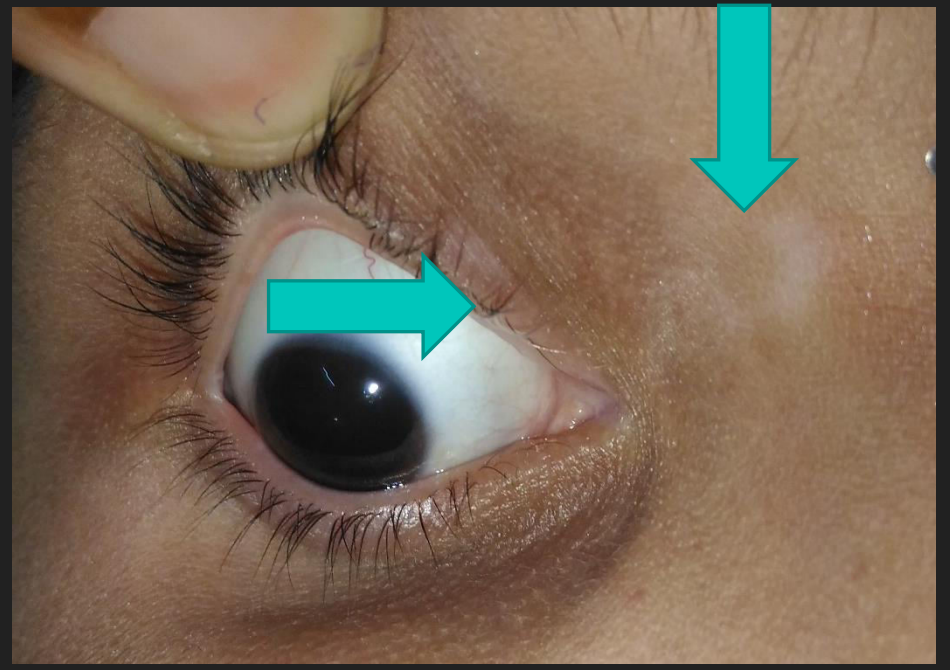






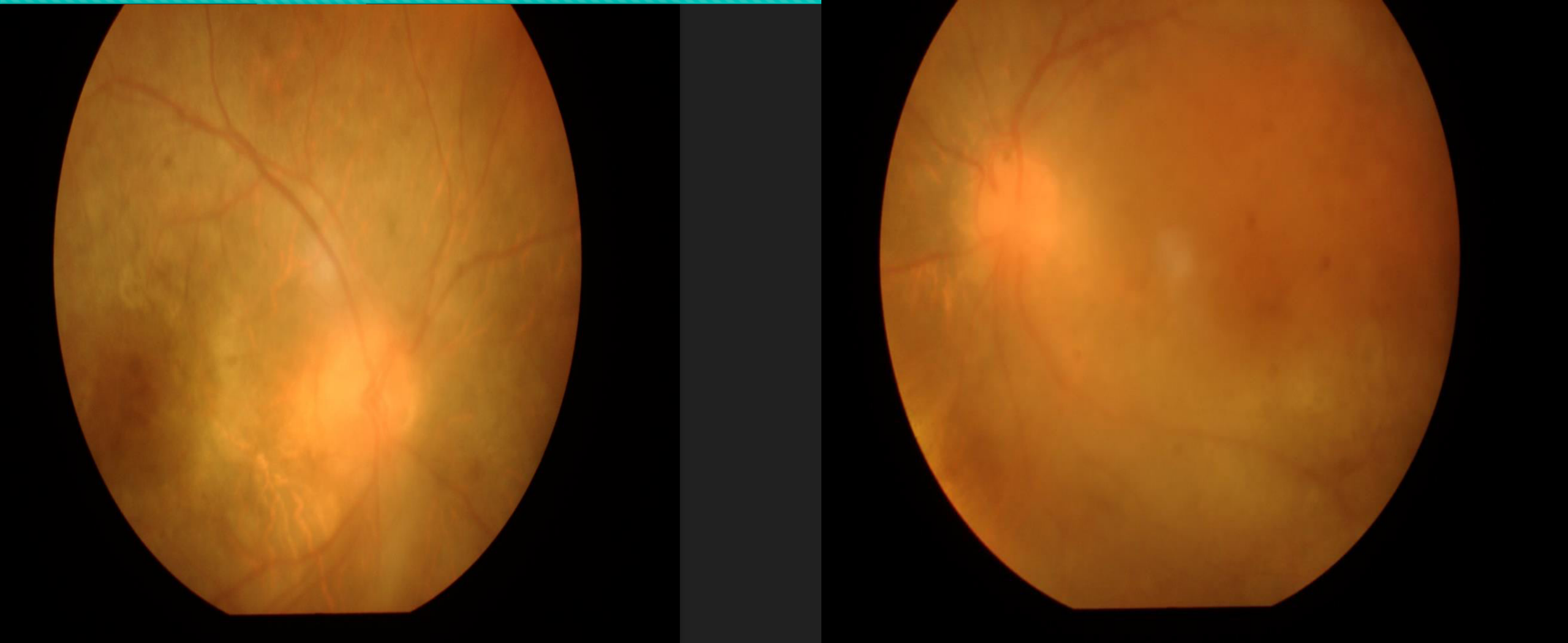
OCULAR FINDINGS

- Uncorrected visual acuity was Finger counting 1 feet and 6/60 of right and left eye respectively.
- Anterior segment examination.
- Poliosis with vitiligo(Suguira's sign),Leukotriachiasis
- No significant refractive error.
- No tinnitus,meningism,alopecia.
- Family History: Younger Sibling has APS-1 and parents have sickle cell trait.





Fundus-Vitritis with sunset glow fundus with exudative retinal detachment with pigmentary changes right eye more affected than left eye



Case 3:

A 13 year old female patient presented
-loss of eyebrows
-eyelashes in both eyes.
-Unaided vision both eyes 6/9.

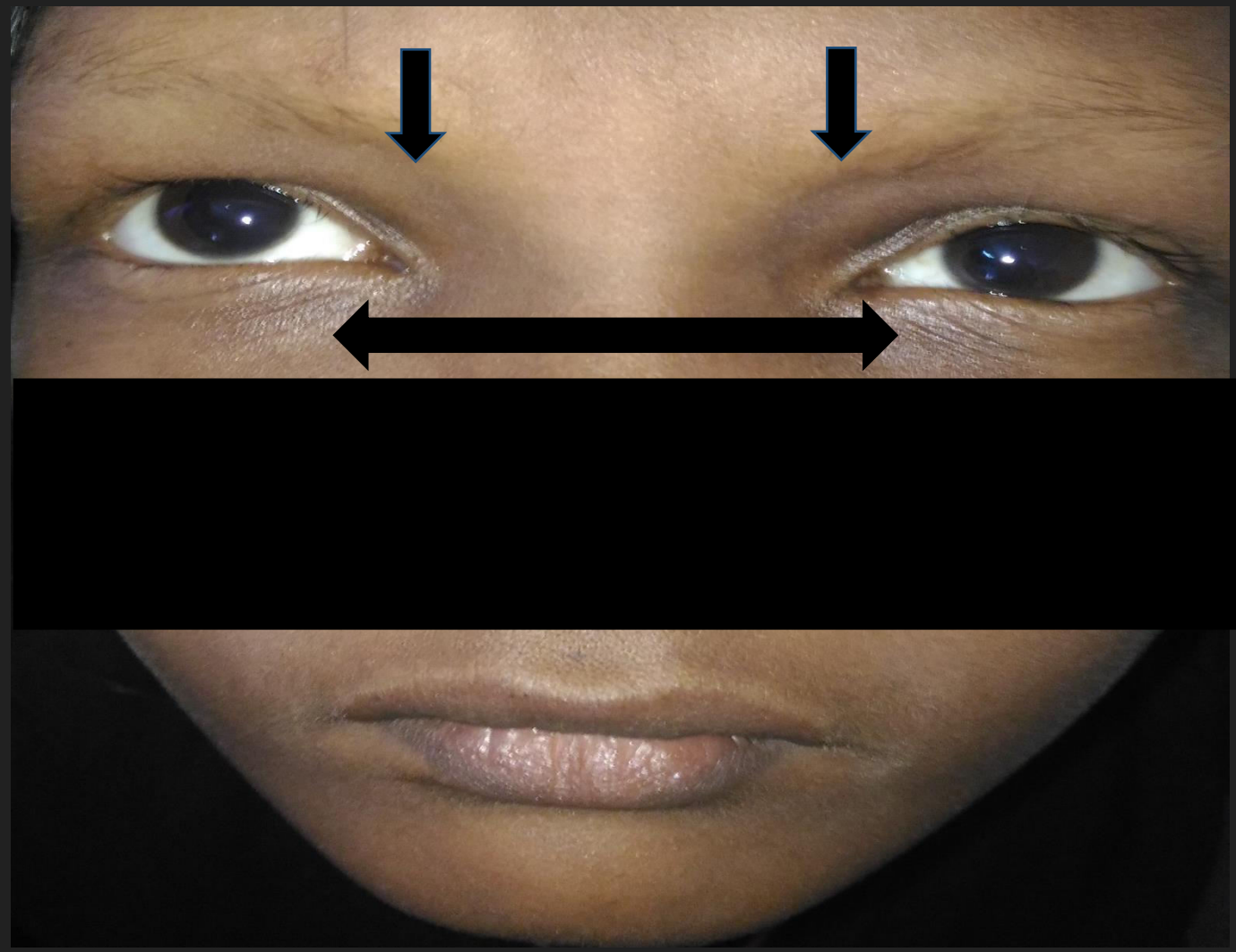
Family History:
Elder sibling has APS-1,Younger sibling has sickle cell anaemia.

Parents have sickle cell trait.

Pupils-Normal

Fundus-Normal

General findings:
Hypoparathyroidism ,adrenal insufficiency,vitiligo



DISCUSSION

DIAGNOSTIC CRITERIA OF (APS) (4)
NEUFELD AND BLIZZARD CLASSIFICATION OF 1980

- **Diagnostic criteria:**

- Presence of two of the major components Chronic mucocutaneous candidiasis, Hypoparathyroidism Adrenal insufficiency

- *INeufeld MB, Blizzard RM. Polyglandular autoimmune diseases.In: Pinchera A, Doniach D, Fenzi GF, Baschieri L, eds. Symposium on autoimmune aspects of endocrine disorders. New York: Academic Press. 1980:357-658.*
- *5. Barker JM, Eisenbarth GS. Autoimmune polyendocrine syndromes.In: Eisenbarth GS, ed. Type 1 Diabetes: Molecular,Cellular, and Clinical Immunology. Denver: Barbara Davis Centerfor Chilhood Diabetes, 2003. Available at: www. barbaradaviscenter.org/ (accessed on 27 February 2009) [Online editionversion 3.0].*

Type 1:	**Chronic candidiasis, hypoparathyroidism, autoimmune adrenal insufficiency**
Type 2	Autoimmune adrenal insufficiency (always present) + autoimmune thyroid disease and/or Type 1 DM
Type 3	Autoimmune thyroid disease + other (excluding Chronic candidiasis, hypoparathyroidism, autoimmune adrenal insufficiency)
Type 4	Two or more organ specific autoimmune diseases (which did not fall into type 1, 2 or 3).

CONCLUSIONS

- APS-1 is a complex syndrome characterized by multiple-organ damage.
- Although rare, it must be diagnosed in early stages, given its high morbidity and mortality. Treatment and follow-up are demanding and require a multidisciplinary approach.
- The main objective is to preserve the patient's quality of life and psychosocial support is essential for most patients.
- Progressive keratopathy and/or retinopathy can lead to severe visual loss and pain in patients with APS1.
- Although no treatment has shown efficacy regarding the APS1-associated ocular manifestations, ophthalmologic examinations are recommended in these patients.