Masquerade Syndrome

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Masquerade syndrome includes a group of malignant and non-malignant systemic or primary ocular disease that clinically present in the eye as an intraocular inflammation or uveitis.

Uveitis Masquerade accounts for 5% of patients with uveitis in a tertiary care centre. An awareness of the presence of and early recognition of the masquerade is of utmost importance as Ocular Masquerade may be the first sign of a life threatening disease.

The various conditions that can manifest in the eye as a masquerade includes:

MALIGNANCIES

Adults:
- Primary Central Nervous System lymphomas
- Primary Intraocular lymphomas
- Systemic NHL Metastatic To The Eye
- Metastatic Carcinoma: Breast / Lung / Renal

Children:
- Leukaemia
- Retinoblastoma
- Medulloepithelioma
- Juvenile Xanthogranuloma

NON-MALIGNANT MASQUERADE

Intraocular foreign body / Retinal Detachment / Retinitis pigmentosa / Pigment Dispersion Syndrome / P. Acnes Infection

Many entities present as chronic intraocular inflammation and a thorough workup to exclude a masquerade syndrome should be carried out in the following situations.

1. All undiagnosed inflammatory disease
2. Intraocular inflammations with atypical clinical features and course
3. Inflammations that do not respond to adequate medical therapy
4. Age <5 years / >50 years

Because of the nature of the underlying disease, which has detrimental consequences, early diagnosis and prompt treatment are critical.

This photoessay is on primary intraocular lymphoma (PIL)

Primary Intraocular Lymphoma is the commonest condition presenting as an ocular masquerade. PIL is a large B cell Non Hodgkin Lymphoma, presenting in the 5th to 7th decade in immunocompetent persons. Presentation in an younger age group is seen in the immunocompromised. This entity is commonly associated with CNS lymphoma and rarely with visceral and nodal lymphoma. This condition is bilateral in 80% of cases. The ocular presentation may be varied and may manifest as vitritis, sub retinal and sub RPE creamy white infiltrates, vasculitis, retinitis or as an uveal mass lesion.

90% of patients with PIL develop CNS Lymphoma while only 5% of patients with CNS Lymphoma will develop intraocular manifestations. Hence a detailed systemic workup, neurological investigations,
like cranial MRI and lumbar puncture are essential. Vitreous biopsy, sub retinal or subRPE aspirates or a retinochoroidotomy may be necessary for histopathological confirmation.

Treatment recommendations depend on whether the lesions are confined to the eye alone both eye and CNS involvement or there is recurrence following primary therapy.

**Treatment recommendations:**

1. **Intraocular Lymphoma alone**
   - XRT to eyes only
   - Systemic Chemotherapy

2. **Intraocular and CNS Lymphoma**
   - XRT to eyes + / - BRAIN
   - Systemic Chemotherapy

3. **Recurrent Intraocular Lymphoma**
   - Intravitreal salvage chemotherapy with Methotrexate, Rituximab, Anti CD-20 Monoclonal antibody. Requires

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**Table 1. Scheme of Investigating a patient with chronic Intraocular Inflammation**

<table>
<thead>
<tr>
<th>Chronic Intraocular Inflammation</th>
<th>B/H</th>
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<tbody>
<tr>
<td>Intermediate Uveitis, Vitritis, Peripheral Retinitis, with or without Ant Uveitis, No Observable Retinal or Choroidal Lesions</td>
<td>PARSPLAN EXUDATE</td>
</tr>
<tr>
<td>SarcoiD</td>
<td>NO</td>
</tr>
<tr>
<td>Toxoplasma</td>
<td>YES</td>
</tr>
<tr>
<td>Toxocara</td>
<td>NEUROLOGICAL SYMPTOMS</td>
</tr>
<tr>
<td>TB</td>
<td>Neuro Workup/Imaging</td>
</tr>
<tr>
<td>R/O PCNSL/MS</td>
<td></td>
</tr>
</tbody>
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**Fig. 1.** a-c : (a) Fundus picture at presentation showing optic nerve infiltration in Primary Intraocular lymphoma. (b) Histopathological evidence of large B cells in vitreous biopsy specimen (c)Fundus photograph comparing the lesion before and after radiotherapy.
multiple intravitreal injection and carry a very high recurrence rate on cessation of therapy.

The first case shows regression after radiotherapy in a biopsy proven case of primary intraocular lymphoma which presented as disc infiltration, disc edema and vitritis. Vitreous biopsy specimen showed large B cells suggestive of PIOL. Note that regression of the lesion is unfortunately associated with optic atrophy as evidenced by the disc pallor and functional loss in this patient. (Fig. 1 a-c)

The second case presented as creamy subretinal infiltrates temporal to the macula in a 55 year old male patient who attended our clinic with complaints of defective vision and floaters. Vitreous biopsy and chorioretinal biopsy specimen showed large B cells suggestive of PIOL. This patient was managed by systemic chemotherapy and showed good resolution of the lesion with residual RPE scarring (Fig 2 a-c).

Fig. 2. a-c. (a) Fundus picture at presentation showing the creamy subretinal infiltrates temporal to the macula in a patient with PIOL. (b)Histopathological confirmation of large B cell infiltrates in the chorioretinal biopsy specimen (c)Comparative fundus pictures before and after systemic chemotherapy showing good resolution and residual RPE scarring