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Candidates are frequently asked to examine the patient’s eyes in the exam as a number of conditions make good cases for the exam and are often readily available. It is therefore important that you have given consideration to the most commonly encountered cases and practised a routine for the examination. You will rarely be expected to perform every aspect of the eye examination, although this is summarised below.

**SUMMARY OF EXAMINATION OF THE EYES**

Always start by checking whether the child can see out of both eyes

**INSPECTION**
- Ptosis, nystagmus, strabismus, cataracts, pupil size inequality
- Structural anomaly: exophthalmos, endophthalmos, eyelid defects, coloboma, craniosynostosis

**PUPILS**

Red reflexes
Pupillary reflexes: direct light reflex, consensual light reflex, accommodation

1. Large pupils:
   - sympathomimetics
   - alcohol
   - Holmes–Adie – large, reacts slowly to light
   - ocular blindness – consensual response in other eye, nil from affected eye
   - cortical blindness – no response to light but reacts to accommodation

2. Small pupils:
   - opiates
   - Horner’s syndrome
   - Argyll Robertson (very rare even in adults)

**VISUAL ACUITY**

Depends on age of child:
- 4 weeks – fix on parent’s face, VEP, Catford Drum, preferential looking tests
- 6 weeks – follow object 90 cm away through 90° (not to midline)
- 3 months – follow object at 90 cm through 180° when supine
- 10 months – picks up raisins with pincer grip, test with each eye covered (may need to cover eye with a patch)
- 1 year – picks up hundreds and thousands
- 2–3 years – miniature toys – use seven known toys and ask ‘What’s this?’; test each eye
- 3 years – Stycar matching letters at 3 m and near
- 5 years – Snellen charts
**EYE MOVEMENTS**

**VISUAL FIELDS**

1. Confrontation perimetry:
   - test each eye separately and then both together to exclude sensory inattention; don’t forget to test for scotomata

2. Defects:
   - concentric decrease – retinitis pigmentosa
   - central scotoma – macular lesions, benign and pathological ICP
   - bitemporal hemianopia – craniopharyngioma
   - homonymous hemianopia – optic tract lesion
   - quadrantanopia – upper: lower fibres in temporal radiation lesion ± speech; lower: upper fibres in parietal radiation

**TEST FOR STRABISMUS**

Cover/uncover test

**FUNDOSCOPY**

Begin with ophthalmoscope at +12 dioptres (red numbers) and gradually adjust it until you can focus on the retina. Look at:

- Cornea – corneal abrasions
- Lens – cataracts
- Disc – optic atrophy, papilloedema, glaucoma
- Arteries – know grading of hypertension
- Retina – exudates, haemorrhages, retinitis pigmentosa

**SHORT CASES FOR EYE CONDITIONS**

**BENIGN INTRACRANIAL HYPERTENSION**

**STEM:** Please examine Sharon’s eyes. She has bad headaches and is sometimes sick in the morning.

**PRESENTATION OF EXAMINATION FINDINGS**

Sharon is an overweight teenage girl with papilloedema on fundoscopy. She has no retinal haemorrhages or changes suggesting hypertensive retinopathy and her blood pressure is normal. Confrontation perimetry is normal but she has bilateral central scotomata. Her vision is a little blurred on distance testing but otherwise the neurological examination is entirely normal.

*Thinking pause*.....

The most likely diagnosis in this setting would be benign intracranial hypertension.
**How would you confirm the diagnosis?**
I would want to perform a CT scan to exclude a space-occupying lesion and hydrocephalus. A lumbar puncture should be performed to measure the CSF pressure.

**What is the cause of BIH?**
There are numerous causes for this including haematological conditions, endocrine causes, drugs, trauma, infections and other systemic conditions, although in 50% of the cases the cause is unknown. The exact pathophysiology is uncertain but the most popular theory is decreased CSF absorption.

**How would you manage this patient?**
BIH is generally a self-limiting condition although various measures can be used to reduce intracranial pressure. Reduction in CSF volume by repeated removal at daily lumbar punctures can be tried but this temporary measure represents an unpleasant ordeal, requiring sedation. Corticosteroid treatment with dexamethasone has been shown in a few reports to reduce pressure but this has not been substantiated. For patients in whom steroids are unsuccessful, acetazolamide can be used either alone or, more commonly, with a loop diuretic. If medical treatment fails a ventriculoperitoneal or lumboperitoneal shunt can be inserted, with good results. Very rarely optic nerve decompression may be required to relieve symptoms.

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**BITEMPORAL HEMIANOPIA**

**STEM:** Stephen’s parents have noticed that he has a tendency to bump into things. They are also concerned that he is the smallest in his class at school. Please examine his eyes.

**PRESENTATION OF EXAMINATION FINDINGS**
Stephen is a 10-year-old boy who appears short and relatively overweight. He has bitemporal hemianopia on visual field testing by confrontation. His discs are pale but there is no papilloedema present. The remainder of the cranial nerves are intact and I would like to do a full neurological examination. I would also like to plot his height and weight on a growth chart appropriate for his age and sex. I would like to ask specifically about symptoms of diabetes insipidus and hypothyroidism although he has no clinical evidence of thyroid disease.

**Thinking pause…..**
Stephen is a boy with bitemporal hemianopia, which may be secondary to a space-occupying lesion compressing the optic chiasma, the most likely cause being a craniopharyngioma.

**How would you confirm your diagnosis?**
Plain lateral skull X-ray is often diagnostic in revealing a calcified mass eroding the clinoid process with an abnormally enlarged sella. The
diagnosis, and any suprasellar extension, can be confirmed by CT or MRI. Assessment of pituitary function will also be necessary.

How is this condition managed?
Surgical resection is indicated if there are visual or neurological disturbances. Between 75 and 80% of patients can have their tumours debulked with a recurrence rate of 20–25%. Steroids can be used preoperatively to reduce pressure and vasopressin used to control diabetes insipidus. Hormone deficiencies should be corrected and hydrocortisone is always required for the stress of surgical procedures even if ACTH levels are normal. Long-term follow-up with CT scanning, endocrine function and vision testing are necessary to monitor the efficacy of treatment.

What is the prognosis for this condition?
Craniopharyngioma should be considered to be a chronic condition. If there is no evidence of disease or calcification on CT scan there is an estimated 70% 10-year survival rate. If residual tumour or calcification remains after surgery, then radiotherapy may be indicated. There is no role for chemotherapy at present.

**PTOSIS**

**STEM:** Emma is concerned that her eyelids are droopy. She is quite anxious and everything is exhausting. Please examine her neurologically.

![Ptosis in myasthenia gravis](image)

*Fig. 6.1  Ptosis in myasthenia gravis.*

**PRESENTATION OF EXAMINATION FINDINGS**

Emma is a 13-year-old girl with bilateral partial ptosis. She has normal pupillary reflexes and eye movements are diminished. On repeated blinking exercises she demonstrates ocular muscle fatiguability with increasing ptosis. She also has reduced facial expression. She has difficulty making
hair-brushing movements and is slow to stand from a crouched position. Climbing stairs is also difficult.

*Thinking pause.....*

Emma demonstrates weakness of ocular muscles and proximal muscle weakness with abnormal muscle fatiguability after repeated activity, suggesting a diagnosis of myasthenia gravis. Due to her age this is likely to be the juvenile form, which is similar to the adult autoimmune type and is associated with high titres or antibody to the acetylcholine receptor.

**How would you confirm the diagnosis?**

Diagnosis is made by observing an improvement following administration of edrophonium (Tensilon). Muscle fatiguability can be demonstrated by stimulating the peripheral nerve using surface electrodes at 4 or 10 Hz.

**How would you manage this patient?**

Medical treatment is with anticholinesterases such as neostigmine or pyridostigmine. In the longer term immunosuppressive therapy with carefully tailored alternate day prednisolone or azathioprine may be required to maintain remission. In the emergency situation plasma exchange may be required for crisis if there is respiratory paralysis or bulbar paralysis. If a thymoma is present or the response to medical therapy is poor, thymectomy may be performed.

**What are the causes of ptosis?**

Causes of ptosis are as outlined in Table 6.1.

<table>
<thead>
<tr>
<th>Type of ptosis</th>
<th>Cause</th>
<th>Related condition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Unilateral</td>
<td>IIIrd nerve palsy complete/incomplete (partial) (fixed dilated pupil, eye down and out)</td>
<td>Posterior communicating artery aneurysm, Suprasellar tumour, Ophthalmoplegic migraine, Orbit lesion, Cavernous sinus thrombosis, Midbrain tumour</td>
</tr>
<tr>
<td>Bilateral</td>
<td>Congenital Myasthenia gravis Myopathy</td>
<td>Wrinkling forehead, fatiguability</td>
</tr>
</tbody>
</table>
**NYSTAGMUS**

**STEM:** Darren has always had jerky eye movements. Please examine his eyes.

**PRESENTATION OF EXAMINATION FINDINGS**

Darren is a 4-year-old boy with bilateral horizontal nystagmus. The intensity of the nystagmus is equal on both sides and is independent of the direction of gaze. He has normal vision. Darren also has normal cerebellar function and the remainder of the neurological examination is normal.

*Thinking pause.....*

In this well boy with no other signs the most likely diagnosis is congenital nystagmus. I would also like to check his hearing and ocular fundi.

**What is ‘congenital’ nystagmus?**

It is isolated nystagmus of unknown cause and is sometimes familial. The condition may improve with age.

**Causes of nystagmus**

Nystagmus describes involuntary oscillations of the eye, which may be horizontal, vertical or rotatory (Table 6.2). It is defined by the fast phase, but it is the slow phase which is pathological, other than pendular nystagmus, where there is no fast phase. Nystagmus may be caused by pathology in the brainstem, cerebellum, cervical cord or inner ear.

<table>
<thead>
<tr>
<th>Table 6.2 Causes of nystagmus</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Type of nystagmus</strong></td>
</tr>
<tr>
<td>-----------------------</td>
</tr>
<tr>
<td>Ocular</td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td>Central</td>
</tr>
<tr>
<td></td>
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<tr>
<td></td>
</tr>
<tr>
<td>Cerebellar</td>
</tr>
<tr>
<td>Vestibular</td>
</tr>
<tr>
<td>Positional</td>
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<td></td>
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<td></td>
</tr>
</tbody>
</table>
STEM: Eilidh is a 6-year-old girl whose parents are worried that there is something wrong with her eyes, particularly after school in the evenings. Please examine her eyes.

PRESENTATION OF EXAMINATION FINDINGS

Eilidh is a 6-year-old girl with misalignment of the visual axis, which is normally corrected by wearing glasses. Her right eye turns inwards at rest when her glasses are removed. The angle subtended by the eyes does not vary with the direction of the gaze. The cover test confirms the presence of a manifest right-sided strabismus. She appears to have impaired near vision with intact distance vision, although more formal testing is required to assess the extent of the refractive error. Her optic fundi are normal.

Thinking pause…..

Eilidh has a concomitant (non-paralytic) manifest squint secondary to a hypermetropic refractive error.

When would you worry about squints in an infant?

Any infant with a fixed squint or any squint persisting beyond 2 months of age should be referred to a specialist paediatric ophthalmologist for further assessment. Although squints are most commonly due to failure to develop binocular vision due to a refractive error, cataracts, retinoblastoma and other intraocular causes must be excluded. Prevention of amblyopia is essential and refractive errors are corrected with glasses.

<table>
<thead>
<tr>
<th>Type of squint</th>
<th>Features</th>
<th>Causes and resulting condition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Non-paralytic</td>
<td>Deviation unchanged</td>
<td>Refractive error: amblyopia, hypermetropia, anisometropia</td>
</tr>
<tr>
<td>(concomitant)</td>
<td>in all directions</td>
<td>Eye disease (often divergent): corneal scar, cataract, optic atrophy,</td>
</tr>
<tr>
<td></td>
<td>Common</td>
<td>retinal disease</td>
</tr>
<tr>
<td></td>
<td>Convergent (85%)</td>
<td>Failure to develop normal binocular vision: usually congenital</td>
</tr>
<tr>
<td></td>
<td>or divergent</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Usually horizontal</td>
<td></td>
</tr>
<tr>
<td></td>
<td>(rarely vertical)</td>
<td></td>
</tr>
<tr>
<td>Paralytic</td>
<td>Deviation varies with direction of gaze</td>
<td>Extraocular muscle palsy: III – divergent squint; IV/VI – convergent</td>
</tr>
<tr>
<td></td>
<td>Rare</td>
<td>squint</td>
</tr>
<tr>
<td>Pseudosquint</td>
<td>Common in children, tends to disappear with</td>
<td>Marked epicanthic folds</td>
</tr>
<tr>
<td></td>
<td>facial development</td>
<td>Small or large interpupillary distance</td>
</tr>
<tr>
<td></td>
<td>Confirmed by negative cover test</td>
<td>Broad nasal bridge</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Facial asymmetry</td>
</tr>
</tbody>
</table>

* Abnormal if more than 6 months old.
Types and causes of squint (abnormal if more than 6 months old) are shown in Table 6.3.

**What are the cover tests?**
There are two types of test used to assess a squint:

1. **Cover/uncover test:** one eye is covered and the other is observed. If the uncovered eye moves to fix upon the object there is a squint, which is present all the time – a manifest squint. Each eye is tested in turn.

2. **Alternate cover test:** if the cover/uncover test is normal, excluding a manifest squint, this test is used. The occluder is moved to and fro between the eyes and if the eye which has been uncovered moves then a latent squint is present.

**LENS DISLOCATION**

**STEM:** Robbie is a 15-year-old boy who is the tallest in his class. Please examine him.

![Fig. 6.2](image_url)

**Fig. 6.2** Lens subluxation. In Marfan’s syndrome the lens is dislocated laterally upwards and outwards.


**PRESENTATION OF EXAMINATION FINDINGS**

Robbie is a tall 15-year-old boy with glasses and a marfinoid appearance. Examination of vision reveals severe myopia and he has upward and outward subluxation of his right lens. There is no evidence of retinal detachment or cataracts, although more formal assessment by a specialist ophthalmologist would be required. In addition he has a high arched palate.
and long thin fingers. The lower segment of his body is longer than the upper segment and his arm span is greater than his height. His joints are hyperextensible.

**Thinking pause…..**

Robbie is a 15-year-old boy with Marfan’s syndrome and subluxation of his right lens. I would also like to examine his chest for evidence of scoliosis and listen to his heart for murmurs suggestive of aortic or mitral valve disease.

**Could the underlying condition be homocystinuria?**

Although both conditions have a similar phenotype, homocystinuria is associated with downward and inward lens subluxation, in contrast to the upward and outward subluxation found in Marfan’s syndrome.

**How is the diagnosis made?**

Diagnosis is essentially clinical, although slit lamp examination and echocardiography are useful. Plasma urinary amino acids can be checked to exclude homocystinuria. Inheritance of Marfan’s syndrome is autosomal dominant and if the diagnosis is uncertain, referral to a clinical geneticist is advisable, when a family tree can be drawn and gene studies undertaken to confirm a gene defect on chromosome 15.

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**OCULOCUTANEOUS ALBINISM**

**STEM:** Jamila has always had jerky eye movements. What do you think may be the cause?

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*Fig. 6.3* A child with oculocutaneous albinism with her parents. (Reproduced with kind permission from Lissauer T, Clayden G. Illustrated Textbook of Paediatrics, 2nd edn. Edinburgh: Mosby, 2001.)
PRESENTATION OF EXAMINATION FINDINGS

Jamila is an 8-year-old girl with depigmentation of the skin, hair and eyes. She lacks pigment in the iris, retina, eyelids and eyebrows. Jamila has pendular nystagmus and photophobia. She wears glasses and her visual acuity is severely impaired.

Thinking pause…..

Jamila is a young girl with oculocutaneous albinism.

What is albinism?

Albinism refers to a group of inherited disorders of the melanin pigment system in which there is a congenital reduction or an absence of melanin formation. Depending on the distribution of depigmentation in the skin and the eye the albinism may be oculocutaneous, ocular or partial. Types of albinism not associated with metabolic disorders include all types of oculocutaneous and ocular albinisms and are autosomal recessive, except for albinism, ocular late onset-sensorineural deafness, X-linked. Types of albinism associated with metabolic defects include the Albinism, oculocutaneous, Hermansky–Pudlak type and Chediak–Higashi syndrome.

All types of albinism are thought to result from different mutations involving the biosynthesis of melanin and are most probably due to an enzyme abnormality. The only enzyme shown to produce albinism when deficient is tyrosinase, which is low or absent in a number of types including tyrosinase negative, minimal pigment type and yellow mutant types of oculocutaneous albinism and in most cases of Hermansky-Pudlak type. However, tyrosinase activity is normal in all other types of oculocutaneous and ocular albinism.

The presence of ocular features is constant and is necessary to make a diagnosis of albinism and is characterised by foveal hypoplasia with an associated reduction in visual acuity that cannot be corrected to normal. Nystagmus is also a constant feature of albinism and usually presents within the first year of life.

How would you manage this patient?

Regular ophthalmological care is essential. Failure to develop the fixation reflex results from lack of eye pigment. There is no treatment but correction of refractive errors and fitting of tinted lenses from early infancy may allow normal fixation to develop. Children are prone to sunburn and skin cancer and protection with sun-hats and sunscreen is essential in bright sunlight. Genetic counselling is also advised.

What is the prognosis for this condition?

This group of disorders is associated with a normal life span except in the case of oculocutaneous albinism, Hermansky-Pudlak type, which is associated with a bleeding diathesis due to storage pool-deficient platelets, and death may result from haemorrhage.
HORNER’S SYNDROME

STEM: Laura has developed a droopy right eyelid. Please examine her eyes and tell us what you find.

Laura is a 2-year-old pale, slim girl with a partial right-sided ptosis. Her right pupil is smaller in diameter than her left pupil but both pupils react to light and accommodation. There is no obvious exophthalmos and her external ocular movements are normal. She has near and distance vision in both eyes but I would like her to have more formal visual testing. She has lost her hair and has an indwelling central venous access device. She has no apical chest scars.

Fig. 6.4 A girl with Horner’s syndrome showing partial ptosis of her right eye with a myotic right pupil.

PRESENTATION OF EXAMINATION FINDINGS

Laura is a 2-year-old pale, slim girl with a partial right-sided ptosis. Her right pupil is smaller in diameter than her left pupil but both pupils react to light and accommodation. There is no obvious exophthalmos and her external ocular movements are normal. She has near and distance vision in both eyes but I would like her to have more formal visual testing. She has lost her hair and has an indwelling central venous access device. She has no apical chest scars.

Thinking pause…..

Laura is a young girl with right-sided Horner’s syndrome.

What is the likely cause of this in her case?

From Laura’s appearance I would diagnose that she is having chemotherapy treatment for an underlying malignancy. I would predict that the site of the tumour is intrathoracic at the upper part of her thorax on the right-hand side, with consequent neuropraxis of the sympathetic distribution to her eye. It is possible that there is a mass higher up the neurological pathway causing compression at the level of the spinal cord, cerebellum or brainstem.

Causes of Horner’s syndrome

- Congenital – heterochromia iridae
- Neuroblastoma with lung apex/cervical sympathetic chain
- Postcardiac surgery – look for thoracic scar
PUPILARY REFLEXES

STEM: Please examine this 4-year-old girl’s eyes.

PRESENTATION OF EXAMINATION FINDINGS

Isla is a 4-year-old girl with pupillary size inequality and abnormal pupillary reflexes. She has no structural anomaly of her eyes and both irises are the same colour. Her left pupil is larger than the right and is unreactive to direct light and consensual light stimulation. The right eye has a normal direct light reflex but it is not possible to elicit a consensual reflex. The left eye has no ocular movements and Isla is blind in her left eye. Ocular movements and vision appear to be normal in her right eye. She did not cooperate for fundoscopy.

Thinking pause…..

Isla is a young girl with a prosthetic left eye.

What may be the underlying diagnosis?
Isla may have been born with a congenital absence of her left eye but the eye socket appears to be normal and able to accommodate a prosthetic eye of comparable size to her other eye. The most likely diagnosis is that she has had retinoblastoma, requiring enucleation of her left eye and subsequent fitting of her prosthetic eye.

INTERNUCLEAR OPHTHALMOPLEGIA

STEM: This 10-year-old boy has difficulty looking to one side. Please examine his eyes.

PRESENTATION OF EXAMINATION FINDINGS

Andrew is a 10-year-old boy with abnormal eye movements. On lateral gaze to the left, the left eye abducts normally while the right eye fails to adduct. Lateral gaze to the right is normal. Visual field testing, pupillary reflexes and fundoscopy are all normal.
**Thinking pause.....**

Andrew has internuclear ophthalmoplegia.

**Can you explain how lateral gaze is normally coordinated?**

The medial longitudinal bundle connects the three ocular nerve nuclei to each other and to other nuclei, including the vestibular nuclei, coordinating the activity of the motor nerves to the eye. The parabducens nucleus, in the pons near to the abducens nucleus, coordinates conjugate lateral gaze. Fibres from here run to the VIth nucleus and to the contralateral IIIrd nerve nucleus via the medial longitudinal bundle. Voluntary gaze to the left is initiated in the right frontal cortex.

**Can you explain this abnormality which is frequently poorly understood by candidates?**

Internuclear ophthalmoplegia (INO) is due to a lesion within the median longitudinal fasciculus. In a right INO there is a lesion of the right median longitudinal fasciculus. On attempted left lateral gaze the right eye fails to adduct. The left eye develops coarse nystagmus in abduction. The site of the lesion is on the side of the impaired adduction, not the nystagmus. Destructive frontal lesions (e.g. tumour or infarct) cause failure of conjugate lateral gaze to the side opposite the lesion. In acute lesions the eyes are often deviated past the midline to the side of the lesion and therefore look *towards the normal limbs*. There is usually contralateral hemiparesis.