Diagnosis and Management of Ocular Motility Disorders

FOURTH EDITION

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This comprehensive, clinically-grounded textbook, now in its fourth edition, supports orthoptists and ophthalmologists in decision-making through the patient care process, from presentation to discharge.

Written by authors with extensive experience in teaching and research, Diagnosis and Management of Ocular Motility Disorders offers a clear and practical overview of assessment and management principles and further explores the clinical features of specific disorders, from amblyopia and infantile strabismus to supranuclear and infranuclear disorders, as well as other miscellaneous disorders of ocular movement. A brand new chapter on congenital cranial dysinnervation disorders reflects recent advances in gene mapping and increased understanding of this condition, and a new appendix provides surgical dose tables for easy reference.

Now in full colour throughout, with additional diagrams and photographs of surgical techniques, this remains the key reference text for orthoptic and ophthalmic professionals managing patients with eye movement disorders.

Praise for previous editions

“The sections on ocular movements, ocular deviation and binocular function are excellent; they are concise, brief, practical and are evidence of the authors’ practical and commonsense approach.”

Eye

“This is a comprehensive text that will appeal to all orthoptists (undergraduates and postgraduates), ophthalmologists and other ophthalmic professions with an interest in binocular vision. It is an essential text for orthoptists as part of their departmental and personal libraries.”

British Orthoptic Journal

WILEY Blackwell
Diagnosis and Management of Ocular Motility Disorders
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Preface to the Fourth Edition

A major change to this fourth edition has been the introduction of coloured illustrations. This has given us the opportunity to provide more diagrams to aid clarity of the text and include photographs of surgical techniques. The expansion of the text and illustrations aims to enlighten both Orthoptists and Ophthalmologists in the decision making for patient care pathways from presentation to discharge.

We have also added two chapters, congenital cranial dysinnervation disorders (CCDD) to reflect the advances in gene mapping and our greater understanding of the aetiology and mechanisms involved in this group of strabismus. The miscellaneous chapter contains details of conditions that both the Orthoptist and Ophthalmologist will encounter in practice but do not fit easily into the other categories. We also include an Appendix, which gives surgical dose tables for easy reference.

As in previous editions new material has been added and evidence base given for new techniques and management choices. Old material has been removed where techniques are no longer used. We have introduced more tables and flow charts to improve clarity and guide the reader through the text.

It is with sadness we have to report that Joyce Mein, the founder of this textbook, passed away and she was greatly missed when compiling this edition. As ever there are many people who require thanks for their contribution, particularly colleagues in Manchester and Sheffield who read chapters and gave helpful comments for improvement. We would especially like to thank Robin Farr for many of the photographs and illustrations, Lawrence Brown for the electrodiagnostic printouts and Alison Ansons for help with the proof reading.
Preface to the First Edition

Our aim in writing this book has been to give guidance on the diagnosis and management of ocular motility disorders, based largely on our own clinical and teaching experience, rather than to provide a comprehensive textbook on this far-ranging subspecialty of ophthalmology. We have emphasized the practical aspect throughout the book. We have attempted to indicate in some detail the problems which may arise in clinical practice and the precautions which can be taken to avoid them, stressing those methods which we have found to be appropriate for each of the conditions discussed. It is hoped that the style we have adopted, using headings, subheadings and tabulation, will make the text easy to read and understand. There is no section on anatomy and physiology. These subjects are referred to only when we have considered more detailed knowledge essential to understanding of the text, as in the chapters on the principles of surgery and on supranuclear disorders. The book is deliberately little illustrated. We have assumed familiarity with standard equipment and have not included pictures of apparatus in most general use. To have provided photographs to illustrate all the ocular motility disorders we discuss would have been a daunting and perhaps impossible task which we reluctantly decided not to undertake. Static photography of what are essentially movement disorders is rarely satisfactory, especially where the more obscure incomitant conditions are concerned. Video-recordings are of greater value, both in diagnosis and in teaching. Neither have we provided a comprehensive list of the numerous excellent publications on all aspects of strabismus: the references we have given are simply intended as a guide to further reading or to the source of relatively new knowledge on some subjects. We confess to a slight bias towards subjects in which we have or have had a particular interest.

The book is divided into three sections. Section I deals in sequence with the assessment of the patient. The questions which it attempts to answer are whether the ocular motility disorder, using the term in its widest sense, is the result of neurological, ocular, muscular or orbital disease; whether defective vision is due to pathological processes or to amblyopia and whether the defect can be treated to achieve a functional cure or, failing that, satisfactory cosmetic improvement. Section II covers in detail the principles and methods of non-surgical treatment, optical, orthoptic and pharmacological, emphasizing practical value and limitations. The principles of surgical management cover relevant aspects of anaesthesia, as well as the pre- and post-operative care of the patient, including the management of complications. Surgical procedures are described, illustrated and evaluated. Botulinum toxin injection as an alternative to muscle surgery is included in this section. Finally, Section III discusses the application of conservative and surgical methods to specific non-paralytic and paralytic conditions and to nystagmus. We have attempted to give an overall
picture of the methods available but have put the emphasis on those which we have found most likely to succeed. We hope we have communicated our conviction that meticulous observation and equally careful recording of the findings are central to the management of strabismus and associated disorders. Detailed descriptions of ocular motility disorders are bedevilled by problems of nomenclature. Universal agreement on terms and their meanings is rarely attained. We have tried to use terminology which is in general use. Alternative or synonymous terms have been added where indicated.

This book could not have been written without the help and advice of our friends and colleagues. Our thanks go to all of them. Our special gratitude goes to Anne Gelder for her admirable secretarial work and for the very many hours she devoted to preparation of the book on our behalf; to Bruce Noble FRCS, Consultant Ophthalmologist at the General Infirmary at Leeds, for his meticulous illustrations in the surgery section, drawn with the informed eye of a surgeon as well as an artist; to Michael Geall, medical photographer and illustrator, for his careful preparation of some illustrations; to Andrew Fox, orthoptic teacher and source of much useful information and to Margaret Harcourt for her patience, support and forbearance.
1 History

Patients with ocular motility disorders present for one or more of the following reasons:

- Manifest strabismus.
- Subjective symptoms.
- Defective ocular movement.
- Nystagmus.
- Abnormal head posture.
- Defective vision.

An accurate and detailed history must be recorded to aid diagnosis, assist in planning management and arrive at a correct prognosis. This first contact with the patient, or the parents if the patient is a child, gives the examiner an opportunity to assess cooperation, to establish a good relationship with the patient and to gain his or her confidence.

**Children**

It is important to involve the child in the history taking as much as possible: surprisingly young children are aware of the questions being asked; they can supply helpful information while allowing the examiner to form an impression of their intelligence and capabilities.

Because so many disorders of vision and ocular motility are associated with developmental anomalies, hereditary disorders or diseases of childhood, the patient must be considered as a whole, whatever the reason for presentation, and a full medical, obstetric, family and social history should be recorded.

**Medical history**

The following information should be obtained:

- the child’s general development, including milestones and progress at nursery or school;
- details of any significant or recent illness and its treatment;
- any ongoing conditions, for example eczema, asthma, etc.;
- any trauma, especially to the head or face;
- any allergies.
Obstetric history

Enquiries should be made about:

- The mother’s health during pregnancy, including the use of prescription medication, recreational drugs and alcohol.
- Whether the type of delivery was spontaneous vaginal and if forceps were used or caesarean section and if there were complications.
- The child’s birth weight and gestational age if known.
- The neonatal history, especially if there were postnatal problems with resuscitation and/or the child was admitted to a special care baby unit.

Family history

Particular reference should be made to a family history of any of the following disorders:

- Strabismus, which is frequently familial. Its presence in other family members makes it more likely that the child has a true rather than an apparent strabismus and should be kept under observation even if the squint is not apparent at the first visit. There may be social problems if more than one child requires treatment. Parents with strabismus or who have children with strabismus may be more reliable witnesses.
- Refractive error.
- Severe visual defects if present in early childhood are frequently hereditary. Parental consanguinity should be noted.
- Several genetic disorders have a high incidence of associated strabismus.

Medical history

The examiner should question the patient on:

- Past and current illnesses.
- Current medication. The drug history is a good guide to the state of the patient’s health and may provide a clue to the cause of an acquired motility disorder. Patients may say that they are in good health but on questioning may admit to being on regular treatment with, for example, insulin or digitalis. Some drugs, notably anticonvulsants, can be the cause of nystagmus.
- Trauma affecting the head or face.
- Other symptoms and signs. Examples are unsteadiness of gait, weight loss or gain, or change in appearance noted by the patient’s family.
- Ophthalmic history. This may include glasses history. In particular, high levels of ametropia or anisometropia indicate that the cause of visual loss may be amblyopia and incorporated prisms that diplopia is likely to be long standing. Other aspects of the history include visual problems, a childhood squint or previous episodes of diplopia.
- Hereditary factors.

Social history

The patient’s occupation should be noted and he or she should be asked how it has been affected by the complaint; patients who hold a driving licence may require advice about continuing to drive. They should be questioned specifically about alcohol consumption, smoking and recreational drugs as these are relevant to recognised motility disorders. Alcoholism can lead to Wernicke’s syndrome, which is a rare condition, and cigarette smoking can result in lung cancer which is associated with the paraneoplastic syndromes.

Adults

Adults are more likely to present with symptoms, particularly diplopia or concern over their appearance. In many cases the symptoms are due to acquired eye movement defects, comprising neurogenic and myogenic palsies, mechanical restriction of ocular movement. It is essential to find and treat the underlying cause if this is unknown. A detailed medical history should be taken in all cases.
**Manifest strabismus**

History taking is discussed below in general terms. Points relating to specific types of strabismus are referred to in subsequent chapters.

**Children**

The majority of patients attending with manifest strabismus are children. Most will have concomitant deviations but children with congenital and acquired defects of ocular movement can present in this way. In all cases it is necessary to ascertain:

- The direction of the strabismus.
- The age at which it was first noticed.
- Who noticed it.
- Whether the onset was sudden or gradual. A history of sudden onset is usually reliable, whereas a gradual onset probably implies a longer duration than that stated. A sudden onset of esotropia should always suggest the possibility of acquired sixth nerve palsy.
- Whether the squint is constant or intermittent. A constant deviation with a variable angle is often mistaken for an intermittent strabismus.
- When the squint is seen, for example on looking up from a book or on lateral gaze. An apparent increase on lateroversion suggests an incomitant deviation but is also a common feature in pseudoesotropia due to epicanthus.
- Whether there has been an increase or a decrease in the angle of deviation since its onset.
- If there are other features that could be related to the strabismus, such as an abnormal head posture. Older children should be asked about symptoms, particularly whether diplopia is present. This is usually but not always indicative of recent onset. Signs suggesting possible diplopia in younger children are:
  1. Increased clumsiness.
  2. Reluctance to play.
  3. Covering or closing one eye, although this is also a common diagnostic sign in intermittent exotropia.

In general, a history of occasional exotropia should be believed, whereas an occasional esotropia may be due to epicanthus or another cause of pseudostrabismus. If the esotropia is not seen on initial examination, risk factors should be considered. A family history of squint and/or hypermetropia, or the presence of even a small esophoria, are all reasons for believing the parent’s observations and keeping the child under review.

**Adults**

Adults may present with a manifest strabismus dating from childhood, either because they would like surgery to improve alignment or because the strabismus has become symptom producing: diplopia may occur if a change in the angle of deviation causes the image to fall outside the suppression scotoma. As much information as possible should be elicited about the type and onset of the strabismus and its subsequent course. The patient should also be questioned about:

- Poor ocular alignment and related psychosocial problems where the patient may complain of difficulty with communication either socially or in the workplace. Mojon-Azzi and Mojon (2009) have shown that headhunters judged that persons with strabismus are perceived as less attractive and less intelligent by potential employers and would find gaining employment more difficult. This was more significant in exotropia and females.
- The reason for attendance. A patient who is embarrassed by his or her appearance may complain instead about minor symptoms. It can be helpful if the possibility of strabismus surgery is suggested.
- Recent change in the angle of deviation. Some patients present with consecutive exotropia and should be asked when this was first noticed. Photographs taken over the past few years may help in this respect.
- The treatment already received. Particular attention should be paid to previous strabismus surgery and the patient’s records should be obtained if possible.

**Defective ocular movement**

Children may present because limitation of ocular movement has been noticed by the parents or another observer. Usually the defective movement is marked, congenital in origin and mechanical or innervational rather than neurogenic: examples are Brown’s syndrome and Duane’s retraction...
syndrome, although the restricted movement in the latter condition is often masked by head movement. Occasionally an overaction secondary to limitation of movement is the presenting sign, for example a marked overaction of an inferior oblique muscle. It is rare for adults to present for these reasons.

The examiner should enquire about:

- the nature of the defective movement;
- how and when it was discovered;
- any associated signs, particularly an abnormal head posture.

**Nystagmus**

**Children**

The parents of an infant or young child may describe rhythmic, irregular or ‘dancing’ eye movements, or the nystagmus may have been detected by a paediatrician or the family doctor.

The parents should be asked:

- The age of onset. The nystagmus may have been present at birth but is more often acquired in infancy.
- Whether they think it is constant or intermittent. They may have noticed an increase in some gaze positions.
- If the movement has remained static or has improved or deteriorated.
- Their opinion of the child’s vision, comparing it with that of siblings at the same age if possible.
- If there are associated signs, such as strabismus, involuntary head movement or an abnormal head posture.
- The obstetric and medical history, particularly whether the child is receiving anticonvulsant therapy, as an overdose of a drug such as phenytoin may induce nystagmus. Older children may present with acquired nystagmus (described later).

**Adults**

Adults with infantile nystagmus occasionally present because their visual acuity is inadequate for their work, hobbies or interests. More usually nystagmus in an adult is acquired, mainly due to brainstem, cerebellar or labyrinthine damage. Acquired nystagmus is frequently associated with oscillopsia and is incapacitating. Other neurological signs are probable. The patient should be questioned about the onset and duration of the nystagmus, with particular reference to all symptoms and signs and to the general medical history. Nystagmus is considered in detail in Chapter 24.

**Abnormal head posture**

A few patients, usually children, may present because of a marked abnormal head posture, adopted to compensate for a congenital ocular muscle palsy, mechanical restriction of ocular movement or infantile nystagmus. Abnormal head posture is discussed in detail in Chapter 2.

**Defective vision**

A common cause of defective visual acuity without outward sign of strabismus or nystagmus is uncorrected refractive error, especially when one eye is principally affected (anisometropic amblyopia). However, defective vision may be the main reason for attendance in a few patients with strabismus and/or nystagmus. Low vision may be the consequence of either condition, or it may be the underlying cause. The possibility of primary ocular pathology makes differentiation very important.

The examiner should enquire into:

- The nature of the defect.
- How it affects the patient.
- Whether poor vision is a subjective complaint or is deduced from the patient’s behaviour. The former applies more to older children and the few adults who present for this reason, whereas the latter applies to young children.
- How the defect was discovered. Monocular low vision in particular is often found through visual screening or by chance.
- Whether vision appears static or is deteriorating.
Any associated symptoms or signs, including headache or strabismus.

The patient’s medical and family history, particularly one of refractive error

**Subjective symptoms**

Symptoms commonly fall into two categories, diplopia and asthenopic symptoms of headache and eyestrain.

**Diplopia**

Diplopia (Table 1.1) occurs when the visual axes are not parallel, causing the image of the fixation object to fall on peripheral retina. The image is seen according to the visual direction of the retinal area stimulated.

Diplopia can be a symptom in the following types of strabismus.

**Concomitant strabismus**

Diplopia is almost invariably horizontal, with uncrossed or homonymous separation of the images in esotropia (i.e. the right-hand image is seen by the right eye) and crossed or heteronymous separation in exotropia. The separation of the images will not alter significantly in different directions of gaze. Diplopia in concomitant strabismus can signify:

- intermittent strabismus;
- recent onset;
- sudden change in the angle of deviation, usually following strabismus surgery;
- marked improvement in the vision of the squinting eye, for example after cataract extraction and lens implant in exotropia secondary to unilateral cataract.

**Decompensated heterophoria and convergence insufficiency**

Diplopia is usually transitory and can be fused by blinking or changing fixation in most cases. The patient may complain of blurred vision

<table>
<thead>
<tr>
<th>Table 1.1</th>
<th>Questions about diplopia</th>
</tr>
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<tbody>
<tr>
<td>Date of onset</td>
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<tr>
<td>Mode of onset</td>
<td></td>
</tr>
<tr>
<td>Sudden</td>
<td></td>
</tr>
<tr>
<td>Gradual</td>
<td></td>
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<tr>
<td>Binocular/monocular</td>
<td></td>
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<tr>
<td>Frequency</td>
<td></td>
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<tr>
<td>Constant</td>
<td></td>
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<tr>
<td>Intermittent</td>
<td></td>
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<tr>
<td>Near or distance</td>
<td></td>
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<tr>
<td>Gaze dependent</td>
<td></td>
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<tr>
<td>With time</td>
<td></td>
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<tr>
<td>Change with time</td>
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<tr>
<td>Worsening</td>
<td></td>
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<tr>
<td>Stable</td>
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<tr>
<td>Improving</td>
<td></td>
</tr>
<tr>
<td>Characteristics</td>
<td></td>
</tr>
<tr>
<td>Horizontal</td>
<td></td>
</tr>
<tr>
<td>Vertical</td>
<td></td>
</tr>
<tr>
<td>Torsional</td>
<td></td>
</tr>
<tr>
<td>Systemic symptoms</td>
<td></td>
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<tr>
<td>At onset</td>
<td></td>
</tr>
<tr>
<td>Brainstem symptoms</td>
<td></td>
</tr>
<tr>
<td>Hemiparesis</td>
<td></td>
</tr>
<tr>
<td>Headache</td>
<td></td>
</tr>
<tr>
<td>Weight loss</td>
<td></td>
</tr>
<tr>
<td>With time</td>
<td></td>
</tr>
<tr>
<td>Facial paraesthesia</td>
<td></td>
</tr>
<tr>
<td>Visual loss</td>
<td></td>
</tr>
<tr>
<td>Orbital pain</td>
<td></td>
</tr>
<tr>
<td>Palpitations, sweating, weight loss</td>
<td></td>
</tr>
<tr>
<td>Generalised weakness, speech and swallowing difficult</td>
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</tbody>
</table>

rather than diplopia if the images overlap. Most patients will be adults, as decompensation in children usually results in persistent manifest strabismus.

**Decompensated congenital ocular muscle palsies**

If decompensation occurs in childhood, the outcome is often manifest strabismus with suppression; however, some children complain of diplopia. When decompensation occurs in adult life, diplopia is generally the presenting symptom.

Vertical muscle palsies are most likely to decompensate, therefore the diplopia is often vertical, with the higher eye seeing the lower image. Torsional diplopia is rarely, if ever, a feature of congenital muscle palsy. The diplopia is usually intermittent
with a gradual onset and the patient is often vague about its duration.

**Acquired limitation of ocular movement**

Diplopia can be horizontal, vertical and/or torsional, depending on the nature of the defect. The separation of the images will decrease in the opposite direction, when fusion may be possible. Diplopia is usually the presenting symptom in children and adults and can be the first sign of serious disease. The onset can be gradual or sudden: patients are usually precise about the onset and the duration. Further information can be found in Chapters 18 to 22.

**Asthenopic symptoms**

Headache and eyestrain can result from decompensating heterophoria and convergence insufficiency, due to the effort required to maintain binocular single vision. Most patients with these complaints will be adults, often those working on exacting visual tasks, recently changed their occupation or activity, or who are unwell or excessively tired. In the context of ocular motor disorders, other causes are uncorrected refractive error, ill-fitting spectacles and accommodative anomalies. When taking the history, the examiner should bear in mind the many other causes of headache, a number of them with more serious implications. Heterophoria and convergence insufficiency are discussed in Chapter 16.

All patients should be asked about:

- The nature of their symptoms.
- When they first became apparent.
- When they occur, with particular reference to variation during the day.
- Whether they have remained static, improved or deteriorated.
- How they affect work and hobbies.
- If they can be overcome in any way.
- Relevant trauma. However, care should be taken in attributing the symptoms to the trauma; the injury may be unrelated or a fall, for example, may be the result of an underlying problem rather than its cause.
- A detailed medical history should be taken, including current medication. The past ophthalmic history should be recorded, with particular reference to childhood strabismus and spectacle wear.

**Reference**

Introduction
Examination of the patient has two main aims:

- Diagnosing the nature and degree of the ocular motor disorder.
- Detecting other ocular and nonocular disorders and assessing their relationship to the motility problem.

The ophthalmic examination should be carried out with strabismus and ocular movement defects in mind. A concomitant strabismus may be the first outward sign of ocular pathology and a constant strictly unilateral esotropia or exotropia in a young child should always be suspect; it is well recognised, for example, that a child with optic nerve glioma can present in this way. A sudden onset of a constant esotropia in a child may be due to a sixth cranial nerve palsy and can be a sign of neurological disease, including cerebral tumour. Infantile nystagmus can result from bilateral ocular pathology, particularly albinism and congenital cataract. When ocular pathology and strabismus coexist, the examiner must assess how much of the vision loss is due to primary pathology and how much to associated deprivation. Additional strabismic amblyopia can develop and may respond to occlusion treatment.

Exotropia can follow loss of vision in one eye in an adult patient and must be considered when planning treatment, for example, in traumatic cataract. The ocular signs can point to the aetiology of acquired cranial nerve palsies. Common examples are those due to diabetes and vascular disease. In some patients defective vision due to ocular pathology can dissociate a heterophoria or a long-standing ocular muscle palsy. Symptoms attributable to decompensation, for example diplopia, may be the reason for presentation in such cases.

Ophthalmic examination should be carried out in a logical sequence, using a cycloplegic agent for fundus examination and refraction at the end of the sequence so that it does not interfere with the performance and interpretation of other tests. Examination is discussed in the following order:

- Assessment of the patient’s appearance, including facial features.
- The presence of an abnormal head posture (AHP).
- Measurement of the uncorrected and corrected visual acuity (VA) at distance and near, assessed after refraction without cycloplegia if necessary.
- Observation of the position and function of the eyelids.
- Observation and measurement of the position of the globe.
Assessment and measurement of the ocular deviation, binocular function and ocular movement.

Examination of the eyes:
1. Analysis of pupil size and reaction.
2. Examination of the anterior and posterior segment.

Additional examination, which may be indicated from the preliminary findings, includes:

- Refraction with cycloplegia.
- Investigation of other aspects of visual function – measurement of the visual fields, colour vision testing, recording the contrast sensitivity threshold and electrodiagnostic assessment.
- General medical examination.
- Laboratory and radiological investigations.

The assessment and measurement of visual function, refraction, ocular deviation, ocular movements and binocular function are referred to only briefly in this chapter because they are discussed in detail in subsequent chapters.

## General appearance

The appearance can provide clues to the patient’s general condition and to the ocular motility disorder.

### Children

#### General features

Many easily recognised conditions are associated with a high incidence of strabismus and eye movement problems. These include:

- hydrocephalus;
- microcephalus;
- albinism;
- Down’s syndrome;
- cerebral palsy;
- craniosynostosis.

#### Facial features

Strabismus can be simulated by:

- epicanthus;
- a narrow or wide interpupillary distance;
- orbital and facial asymmetry;
- a positive or negative angle kappa.

Disorders of the facial skeleton, such as hypertelorism or the craniosynostosis such as Crouzon’s syndrome, are associated with strabismus and, in the latter case, with impaired vision. Facial asymmetry may be a consequence of the branchial arch syndromes (Table 2.1); it can be associated with a head tilt of ocular or nonocular origin. Loss of facial expression is characteristic of Möbius’ syndrome. Both parents’ appearance should also be noted since some disorders are familial.

#### Table 2.1 Branchial or pharyngeal arch syndromes

<table>
<thead>
<tr>
<th>Symmetrical</th>
<th>Asymmetrical</th>
</tr>
</thead>
<tbody>
<tr>
<td>Goldenhar’s syndrome</td>
<td>Microtia and atresia</td>
</tr>
<tr>
<td>Treacher Collins syndrome</td>
<td>Hemifacial microsomia</td>
</tr>
<tr>
<td>Nager’s syndrome</td>
<td></td>
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</tbody>
</table>

### Adults

#### General features

Signs of congenital abnormality can be present, as described above. Significant acquired neurological signs which are easily seen include:

- facial nerve palsy;
- tremor;
- ataxia;
- deafness (unilateral deafness may be due to an acoustic nerve tumour).

The main signs suggestive of Graves’ orbitopathy are lid retraction and proptosis in thyrotoxicosis.

#### Facial features

Loss of facial expression suggests the possibility of myasthenia, muscular dystrophy or chronic progressive external ophthalmoplegia. Outward evidence of facial trauma can indicate the possible nature of defective ocular movement, for example:

- Flattening of the zygomatic arch can be associated with a blow-out fracture of the orbital floor.
• Scarring in the region of the trochlea, often from windscreen glass, can result in an acquired Brown’s syndrome or less commonly a superior oblique palsy.

### Head posture

An AHP should be assumed to result from an ocular cause until proven otherwise. It may result from nonocular causes or may compensate for an ocular condition. The underlying condition can be congenital or acquired. The head posture can be observed from approximately six months of age, when an infant can normally sit unsupported. An AHP may be present all the time or adopted only when needed, especially if it is uncomfortable to maintain.

### Components

An AHP has three possible components, which may be present singly or in combination:

- head tilt to the right or left shoulder;
- head turn to the right or left side;
- head elevation or depression.

### Examination

Initially nonocular and ocular head postures must be differentiated. The essential differences between them are:

1. The presence or absence of a strabismus or other ocular abnormality, for example nystagmus, when the head is straightened.
2. The ability to straighten the head. It should always be possible to straighten the head if the cause is ocular but some patients with nonocular head postures may be unable to do so because of bony changes in the cervical spine or muscle contracture.

- The components of the head posture should be analysed, noting the position in which the eyes are placed. Straightening the head, then allowing it to resume a comfortable position, often simplifies recognition of its different components.
- The head posture should be assessed for near and distance as it may vary significantly from one distance to the other; for example, a patient with a partial sixth nerve palsy may need a head turn only for distance.
- Long-term observation may be needed to see an intermittent head posture. Observation of a child when playing may reveal, for example, the head elevation seen in esotropia with the ‘A’ pattern, which may be too uncomfortable to maintain all the time.
- A cover test should be performed with the AHP and with the head straight, noting the difference in the degree of ocular deviation and whether there is recovery to binocular single vision (BSV).
- The head posture should be observed when testing the monocular and binocular VA. In some patients it is present only when clear vision is required.
- Early unposed photographs can indicate the duration of the head posture. They should show whether it is consistent and if it has changed with time. An intermittent AHP may be seen on photographs.

The investigation is summarised in Flow 2.1.

### Causes of abnormal head posture

(Flow 2.2)

#### Nonocular head postures

**Congenital**

**Nonocular torticollis (congenital muscular torticollis)**

In a series by Cheng et al. (2000) just under 50% had a sternomastoid tumour, about a third had a tight sternomastoid muscle but no tumour and the remaining 20% had neither a tumour nor tightness of the muscle (postural torticollis). The typical head posture comprises a head tilt to the affected side, a head turn to the normal side and head elevation.
Differential diagnosis is made by:

- Attempting to straighten the head, which will be difficult in nonocular torticollis, but easily achieved in ocular torticollis.
- Examination of the ocular posture by cover test. There should be no manifest deviation in nonocular torticollis, but a heterotropia or nystagmus should be detected in ocular torticollis when the head is straight.

**Acquired**

There are several possible acquired causes of nonocular AHP:

- Deafness. If this is unilateral, or is more marked in one ear, the patient may turn his or her head to the more affected side. This is a common cause of head turn, especially in the elderly. It is worth questioning the patient about hearing
if there is an unexplained or atypical head posture.

- Defects of vestibular function. The ocular tilt reaction consists of a cyclovertical strabismus (skew deviation), ocular torsion and a head tilt towards the hypotropic eye (see Chapter 23).
- Disorders of the cervical spine.
- Habit. Although this is commonly cited as a cause of AHP by parents, it is unlikely to explain a persistent abnormality and other reasons for the head posture should be sought.

Abnormal head posture may be associated with other less common neurological or medical conditions, including brain tumours, stressing the need for thorough investigation (Boutros and Al-Mateen 1995).

Ocular head postures

The main ocular causes for an AHP are vertical and horizontal incomitance and nystagmus (Kushner 1979). The reasons for adopting the head posture are:

- To obtain or maintain a field of BSV and place it centrally. This applies to:
  1. All forms of congenital or acquired paralytic strabismus, including those due to innervational and mechanical restriction of movement.
  2. Alphabet patterns.
  3. A few cases of heterophoria and convergence deficiency.
  4. High degrees of anisomyopia (the ‘heavy eye’ syndrome).
- To obtain better VA and/or centralise the visual field. This applies to:
  1. Nystagmus with an eccentric null area in which the oscillations are much reduced or absent.
  2. Gross restriction of ocular movement, which prevents foveal fixation in the primary position.
  3. Visual field defects. An early onset homonymous hemianopia can be associated with a head turn towards the side of the visual field defect (Paysse and Coats 1997).
  4. Ptosis.
A head tilt is seen in patients with dissociated vertical deviation but the reason for the tilt is not clear. Abnormal head postures in the ocular conditions referred to above are discussed in the relevant sections of this book.

**Head movement**

The patient should be observed for abnormalities of head movement. These can take the following forms:

- **Movement of the head rather than the eyes in order to maintain BSV.** This is often seen in Duane’s retraction syndrome, when it can successfully mask the limitation of ocular movement. This sign may be the observer’s first indication of the syndrome. Patients with chronic progressive ophthalmoplegia also substitute head movement for eye movement.

- **Head nodding or head shaking** is a feature of some types of nystagmus and is commonly seen in spasmus nutans in which the rate of head movement does appear to compensate for the nystagmus.

- **Exaggerated head thrusts on attempting to change fixation,** which are seen in congenital ocular motor apraxia.

**Visual acuity**

Visual acuity should be measured monocularly for near and distance, with and without spectacles if worn. If monocular vision is defective, it is advisable to test the acuity with both eyes open. A pinhole can be used to assess how much of the defect can be accounted for by an uncorrected refractive error. Care should be taken when interpreting the result of pinhole vision as some patients find it difficult to use the pinhole and all patients should be refracted. Any special features should be noted, such as ‘screwing up’ of the eyes, an AHP or head movement. When vision is defective the patient’s optimum reading distance and near VA should be recorded in addition to the near vision.

Refraction without a cycloplegic agent may be useful at this stage, both to find the best corrected VA and to ascertain the type and degree of refractive error, which can have a bearing on further investigation as well as on diagnosis.

The assessment of vision may require modification, depending on age and cooperation. Methods suitable for young children are described in Chapter 4.

**Eyelids**

**Palpebral fissures**

Variants from the normal palpebral fissure shape or size can result in pseudostrabismus or can be characteristic of certain ocular motility disorders.

**Shape**

Mongoloid palpebral fissures are typical of Down’s syndrome, which has a high incidence of esotropia. Urrets-Zavalia et al. (1961) stated that the type of inferior oblique dysfunction influenced the alphabet pattern:

- Mongoloid fissures favoured the production of A-pattern esotropia and V-pattern exotropia.
- Antimongoloid fissures favoured the production of A-pattern exotropia and V-pattern esotropia (see Chapter 17).

**Fissure changes during ocular movement**

An increase or decrease in the height of the palpebral fissure can be characteristic of some incomitant conditions, for example:

- Narrowing of the fissure on adduction and widening on abduction are diagnostic features of Duane’s retraction syndrome.
- The fissure commonly widens on attempted elevation in Graves’ orbitopathy and orbital blow-out fracture.
- Widening of the fissure occurs on abduction in some cases of acquired sixth cranial nerve palsy and on adduction in third nerve palsy associated with aberrant regeneration.

**Congenital anomalies**

**Epicanthus**

Epicanthus is a fold of skin that arises in the medial portion of the upper eyelid and is inserted into the
lower eyelid at the medial canthus. It is usually bilateral and symmetrical. It causes an apparent increase in the intercanthal distance and therefore shortens the palpebral fissure along its horizontal axis.

Epicanthus gives rise to pseudoesotropia and is the most common cause of this condition in young children. It can also accentuate an existing esotropia. Epicanthus is physiological and usually disappears by 7 or 8 years of age in Caucasians, as the skin fold is gradually taken up by the developing nasal bridge. If it persists and is marked, oculoplastic surgery may be required.

Blepharophimosis

The features of this condition are:

- marked epicanthic folds;
- partial ptosis;
- epicanthus inversus, which is a much less common form of epicanthus in which the skin fold arises in the lower eyelid and is inserted into the upper eyelid;
- telecanthus, which is a widening of the intercanthal distance.

As a result of these features, the palpebral apertures are slit-like and unsightly. The condition can be improved by oculoplastic surgery. Both epicanthus inversus and telecanthus can present without blepharophimosis.

Epiblepharon

This anomaly is characterised by a fold of skin running horizontally in the lower eyelid close to the lash line, usually in both eyes. Occasionally the skin can push against the lower eyelid, causing the eyelashes to come in contact with the cornea, but as the lashes are still soft, little irritation results and surgical correction is not required. The fold of skin usually disappears spontaneously by 12 months of age.

Eyelid position and movement

Abnormalities of eyelid position and eyelid movement are commonly associated with ocular movement disorders and should be carefully noted. Correct assessment of the anomaly can aid the diagnosis of the movement disorder and can indicate its aetiology.

Normal position and movement

The eyelid position is symmetrical on both sides. The height of each palpebral fissure is 9–11 mm and the resting position of the upper eyelid margin is 1–2 mm below the superior limbus. This relationship is normally maintained in the different positions of gaze. Movement of the upper eyelid margin from down-gaze to up-gaze measures 15–18 mm.

Eyelid anomalies

Eyelid anomalies comprise:

- upper eyelid too low: ptosis or pseudoptosis;
- upper eyelid too high: lid retraction;
- lower eyelid anomalies;
- abnormal eyelid movement.

Ptosis

Upper eyelid ptosis can be due to:

- underaction of the levator muscle;
- underaction of Müller’s muscle;
- mechanical factors.

Examination aims to establish:

- The degree of ptosis and whether it is unilateral or bilateral.
- Whether it is an isolated defect or is associated with defective ocular movement or other anomaly.
- The reason for ptosis.
- Its effect on VA and binocular vision. Ptosis may be complete, when the upper eyelid obscures the pupil, or partial, when some eyelid movement is possible. If the pupil is obscured by the ptosis during childhood, there is a high risk of stimulus-deprivation amblyopia.
- The size and reaction of the pupil.
Causes of upper eyelid ptosis (Flow 2.3)

Congenital ptosis

Ptosis dating from birth or with very early onset can be unilateral or bilateral.

Features

Complete unilateral ptosis is associated with:

- severe stimulus-deprivation amblyopia;
- myopia or myopic astigmatism in the affected eye in some cases;
- congenital third nerve palsy in a few cases;
- a constant strabismus in the affected eye, either secondary to low vision or as an exotropia resulting from a third nerve palsy.

The earliest possible surgical treatment of the ptosis is essential to prevent further stimulus deprivation and to allow treatment of the amblyopia.

Partial ptosis may be bilateral or unilateral and can be associated with the following conditions:

- A partial third nerve palsy, affecting either all the extraocular muscles supplied by the nerve or only the superior rectus and the levator muscles, which are supplied by its superior division. It is believed that the latter is more common because the two muscles arise from the same embryonic mass.
- A double elevator palsy, in which a true or pseudo ptosis can be associated with a unilateral failure of elevation and hypotropia.
- A congenital Horner’s syndrome, in which slight ptosis results from underaction of the smooth Müller’s muscle. This condition is far more commonly acquired and is described below.
- The Marcus Gunn phenomenon, in which there is a partial ptosis in the primary position which increases or decreases with movement of the lower jaw (see below).
An abnormal head posture of head elevation may be adopted to facilitate vision when the ptosis is bilateral or affects the fixing eye. If the ptosis covers the pupil or there is a manifest strabismus, amblyopia will be present.

**Acquired ptosis**

**Involutional (age-related) ptosis**

Weakness of the levator aponeurosis can develop with age, causing the upper eyelid to droop. A higher than normal skin crease can be seen. Involutional ptosis is usually bilateral but not necessarily symmetrical. Ocular movement is usually normal for age; it is recognised that elevation of the eyes is reduced in the elderly.

**Neurogenic ptosis**

Third cranial nerve palsy involves damage to the third cranial nerve and results in paralysis or paresis of the levator muscle and of all or some of the extraocular and intraocular muscles supplied by the nerve. The degree of ptosis depends on the extent of nerve damage.

Other neurological signs may be present, indicating further investigation. A sudden onset of third nerve palsy with pupil involvement, associated with headache, can be due to aneurysm on the circle of Willis, which may rupture, and is a neurosurgical emergency.

Lid function may improve with time, but if the palsy was due to trauma or an aneurysm in particular, reinnervation of the levator is quite often aberrant (the misdirection syndrome; see below).

Horner’s syndrome involves damage to the sympathetic nerve supply to the eye and results in the following signs:

- Slight unilateral ptosis.
- Miosis of the affected eye.
- Ipsilateral anhidrosis, resulting in warm, dry, flushed skin on the affected side of the face. This is not always present.
- Heterochromia iridis, which is sometimes present in rare congenital cases of Horner’s syndrome.
- There is no associated ocular motor defect other than the ptosis.

A lesion can affect the sympathetic nerve supply at any point along its lengthy pathway from the hypothalamus. Pharmacological testing can help localise the site of the damage. Lesions are most likely to occur in the sympathetic chain, where apical lung tumours are a common cause. If the lesion occurs above the level of the neck, for example, to carotid aneurysm, the third nerve can also be affected, but because of sympathetic damage, the pupil will be small. The examiner should be careful not to misinterpret this sign by assuming that absence of the usual pupil dilation rules out an aneurysm as the cause of the palsy.

Cyclic oculomotor palsy occurs in childhood and is characterised by unilateral ptosis and limitation of ocular movement during its paretic phase and by lid retraction during its spastic phase (see Chapter 19). Ptosis is the usual reason for ophthalmic referral.

**Myogenic ptosis**

Ptosis can be due to disease processes directly affecting the levator muscle:

- Myasthenia. The ptosis associated with this disease is usually partial. It may be unilateral or bilateral and is essentially variable, increasing with fatigue and becoming more marked at the end of the day. There is often limitation of ocular movement and although diplopia is frequently the presenting symptom, myasthenia can present in a variety of other ways.
- Chronic progressive external ophthalmoplegia affecting both eyes is associated with a long history of slowly increasing ptosis.

Myogenic conditions are discussed in Chapter 20.

**Mechanical ptosis**

Mechanical ptosis is usually due to excessive weight of the upper eyelid, caused by oedema, haematoma or tumour. The ptosis is usually unilateral and is partial in the primary position, but may decrease on down-gaze. Trauma is a common cause and the contour of the eyelid can be distorted.

**Pseudoptosis**

In pseudoptosis there is drooping of the upper eyelid on clinical inspection but demonstrably normal eyelid function. The causes are:

- Enophthalmos.
- Retraction of the globe in one or more positions of gaze. Duane’s retraction syndrome is the main example.
Hypotropia. The relationship between the upper eyelid margin and the limbus is normal for the down-gaze position, as can be seen when the hypotropic eye is made to fixate in the primary position and the ‘ptosis’ disappears. Eyes unable to fix in the primary position may mask the full extent of any true ptosis (see below under ‘Inferior rectus tethering’).

Upper eyelid retraction
Retraction is due to overaction of Müller’s muscle, overaction or contracture of the levator muscle or mechanical factors involving the eyelid or ocular movement.

Thyrotoxicosis
This is the most common cause of lid retraction. Both eyes are usually affected, although often asymmetrically; occasionally the retraction is strictly unilateral. Smooth muscle is believed to be unduly sensitive to sympathetic hormones in thyrotoxicosis, leading to overaction of Müller’s muscle. This can result in very variable lid retraction and can also contribute to the lid-lag, which is a common feature of this disease (von Graefe’s sign). Phenylephrine 2.5% drops instilled into the eye blocks the overaction of Muller’s muscle and temporarily corrects the retraction.

Inferior rectus tethering
Mechanical restriction of elevation due to tethering of the inferior rectus in Graves’ orbitopathy results in increased drive to the superior rectus in an effort to elevate the eye. As the superior rectus and levator muscles are synergists, equal innervation goes to the levator, resulting in overaction and lid retraction.

Levator shortening
The levator muscle can become contracted in the cicatricial phase of Graves’ orbitopathy. This may result in impaired lid closure in addition to lid retraction.

Ptosis of the contralateral eye
When there is a moderate degree of unilateral ptosis, an attempt to raise the affected eyelid is accompanied by synergistic overaction of the contralateral levator muscle (Hering’s law), leading to lid retraction of the unaffected eye. This can be particularly obvious when the ptosis is on the side of the preferred eye.

Midbrain disease
Lesions in the upper part of the midbrain can cause unilateral or bilateral lid retraction (Collier’s sign). This is commonly associated with Parinaud’s syndrome.

Postoperative lid retraction
Lid retraction can follow:
- Over-liberal ptosis surgery. Lid closure is also likely to be affected.
- Over-liberal recession of the superior rectus muscle. The anatomical connections between this muscle and the upper eyelid result in retraction.

Lower eyelid anomalies

Lower eyelid ptosis
Lower eyelid ptosis can be caused by excessive resection of the inferior rectus muscle.

Lower eyelid retraction
Lower eyelid retraction is frequently seen in Graves’ orbitopathy and is seen infrequently following a recession of the inferior rectus muscle with or without full division of its palpebral attachments.

Abnormal eyelid movement

Marcus Gunn jaw-winking phenomenon
The jaw-winking phenomenon is a congenital condition in which movement of the jaw results in a change in eyelid position. It is nearly always unilateral.
Features

The features are:

- There is partial ptosis in the primary position.
- The degree of ptosis increases when the mouth is opened and when the jaw is moved, usually to the affected side. The ptosis then decreases on jaw movement to the sound side, when lid retraction is apparent in some cases. In a few patients the converse occurs, with increased ptosis on jaw movement to the sound side. Rapid change in eyelid position occurs on chewing or sucking, and for this reason the anomaly is often detected early in infancy.
- In a minority of patients there is limitation of upward movement of the eye on the affected side, resulting in a hypophoria or, more rarely, a hypotropia. Superior rectus palsy is found in 25% of cases and double elevator palsy is found in another 25% of cases, while anisometropia occurs in 5–25% of patients and amblyopia secondary to strabismus or anisometropia occurs in 30–60% of patients; rarely is it the result of deprivation by a ptotic eyelid (Pratt et al. 1984). The phenomenon is believed to be due to a congenital misdirection of the nerve supply to the levator muscle (superior division of the third cranial nerve) and the pterygoid muscles (mandibular division of the fifth cranial nerve). Lyness et al. (1988) examined the levator palpebrae superioris in 12 patients with the Marcus Gunn phenomenon. They considered that the histological appearance of fibre loss with atrophy and hypertrophy of the remaining muscle fibres was compatible with neurogenic atrophy combined with aberrant reinnervation. Similar changes were also found on the clinically normal side, which they suggested might indicate that the causative lesion was located within the central nervous system.

Many patients learn to disguise the wink in time. The severity of the condition varies greatly. If it is marked, Collin (1984) advises bilateral disinsertion of the levator muscles and bilateral brow suspension.

Aberrant regeneration of the third cranial nerve (misdirection syndrome)

The regrowth of nerve fibres may be misdirected following a third nerve palsy, especially if it is due either to trauma or an aneurysm on the circle of Willis. Misdirection causes elevation of the upper eyelid on down-gaze (pseudo-von Graefe’s sign), particularly when the eye is adducted, and can also result in anomalous ocular movement and pupil reaction. Misdirection does not occur in diabetic third nerve palsy. The syndrome is described in detail in Chapter 19.

Facial nerve palsy

Upper or lower motor neurone seventh cranial nerve palsy results in failure of lid closure on the affected side, most noticeable when the patient blinks. The eye elevates if Bell’s phenomenon is present, exposing the sclera. The loss of orbicularis tone can also result in ectropion.

Orbicularis tone can be assessed by first asking the patient to close his eyes and keep them closed; the examiner then attempts to raise the upper eyelid, comparing the results in each eye. Slight orbicularis weakness can be detected in this way.

Lid-lag

Lid-lag is characterised by failure of the upper eyelid to follow the downward movement of the globe. This condition commonly occurs in patients with Graves’ orbitopathy, when it is usually bilateral, possibly asymmetrical. It can also result from trauma to the upper eyelid and a levator resection to correct ptosis.

Examination of the eyelids

Eyelid position

The patient should be on eye level with the examiner and should look straight at him or her. The eyelid positions should be compared. If the relationship of the lid margin to the limbus appears abnormal, or if asymmetry is present, the height of each palpebral fissure should be measured with a ruler. The marginal reflex distance (MRD) is the distance between the eyelid margin and the centre of the corneal reflex (Figure 2.1). The comparative height of the eyebrows should be observed: brow wrinkling due to overaction of the frontalis muscle should be noted and may indicate the presence of ptosis.
The upper eyelid skin crease should be measured and compared in each eye.

Partial ptosis can result in pseudohypertropia or it can be associated with a strabismus, commonly hypotropia. The eye position in the primary position should be noted. A cover test should show whether or not a true strabismus is present in patients with ptosis or lid retraction.

Eyelid movement

Anomalous eyelid movement may be apparent from observation of the patient when taking the history and on examination of ocular movement.

- The eyelids should be observed as the eyes move into the main positions of gaze, looking for abnormalities of ocular movement as well as eyelid movement. It is important not to raise the upper eyelids when testing on down-gaze, at least initially, otherwise anomalies such as lid-lag and misdirection of nerve fibres can be missed.
- Eyelid closure should be tested. This can be impaired even if ptosis or lid retraction are present, particularly if these are mechanical or myogenic.

Assessment of ptosis (Table 2.2)

- Initially true ptosis must be differentiated from pseudoptosis. This is usually easily achieved by observation of enophthalmos and retraction of the globe and by a cover test to ensure fixation with the suspected eye.
- The examiner must decide whether a patient with a small degree of upper eyelid asymmetry has mild ptosis of one eye or lid retraction in the other eye. Clues may be obtained from other signs. The presence of ptosis can be confirmed by comparing the marginal reflex distance in each eye. A reduction or posterior extension in the upper eyelid skin crease or slight frontalis muscle overaction may also provide useful clues. Unilateral miosis suggests ptosis due to Horner’s syndrome. Patients with lid retraction may also show lid-lag or other signs of Graves’ orbitopathy. Ptosis on one side can result in lid retraction on the other side, as described above, but the ptosis in these cases is usually more obvious.
- Asymmetric bilateral ptosis. If bilateral ptosis affects one eye more than the other, as seen, for example, in some patients with myasthenia, it can be mistaken for unilateral ptosis. The additional innervation required by the levator muscle to elevate the more ptotic eyelid goes equally to the levator in the less affected eye, masking the ptosis or even resulting in apparent lid retraction. The ptosis is made apparent either by covering the more affected eye or by holding up the more ptotic lid.
- Levator function. This should be assessed by preventing the action of the frontalis muscle. The patient first closes his eyes and the examiner holds the eyebrow against the frontal bone. This excludes frontalis contraction when the patient opens his eyes; elevation of the upper eyelid is then due to residual action in the levator and Müller’s muscles. Levator function can be measured by recording the amount of excursion of the upper eyelid from down-gaze to up-gaze. The degree of levator function is of great importance in planning ptosis surgery.
- Abnormal head posture. Elevation of the head is the common AHP, adopted to obtain foveal fixation. If the ptosis is unilateral and is associated with limited upward eye movement, there may also be a head tilt to the affected side to maintain BSV. The degree of head posture...