check

Independent learning program for GPs

Unit 577
November 2020

Paediatric health

RACGP

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Published by

The Royal Australian College of General Practitioners Ltd
100 Wellington Parade
East Melbourne, Victoria 3002, Australia

Telephone 03 8699 0414
Facsimile 03 8699 0400
www.racgp.org.au

ABN 34 000 223 807
ISSN 0812-9630

We acknowledge the Traditional Custodians of the lands and seas on which we work and live, and pay our respects to Elders, past, present and future.

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Paediatric health

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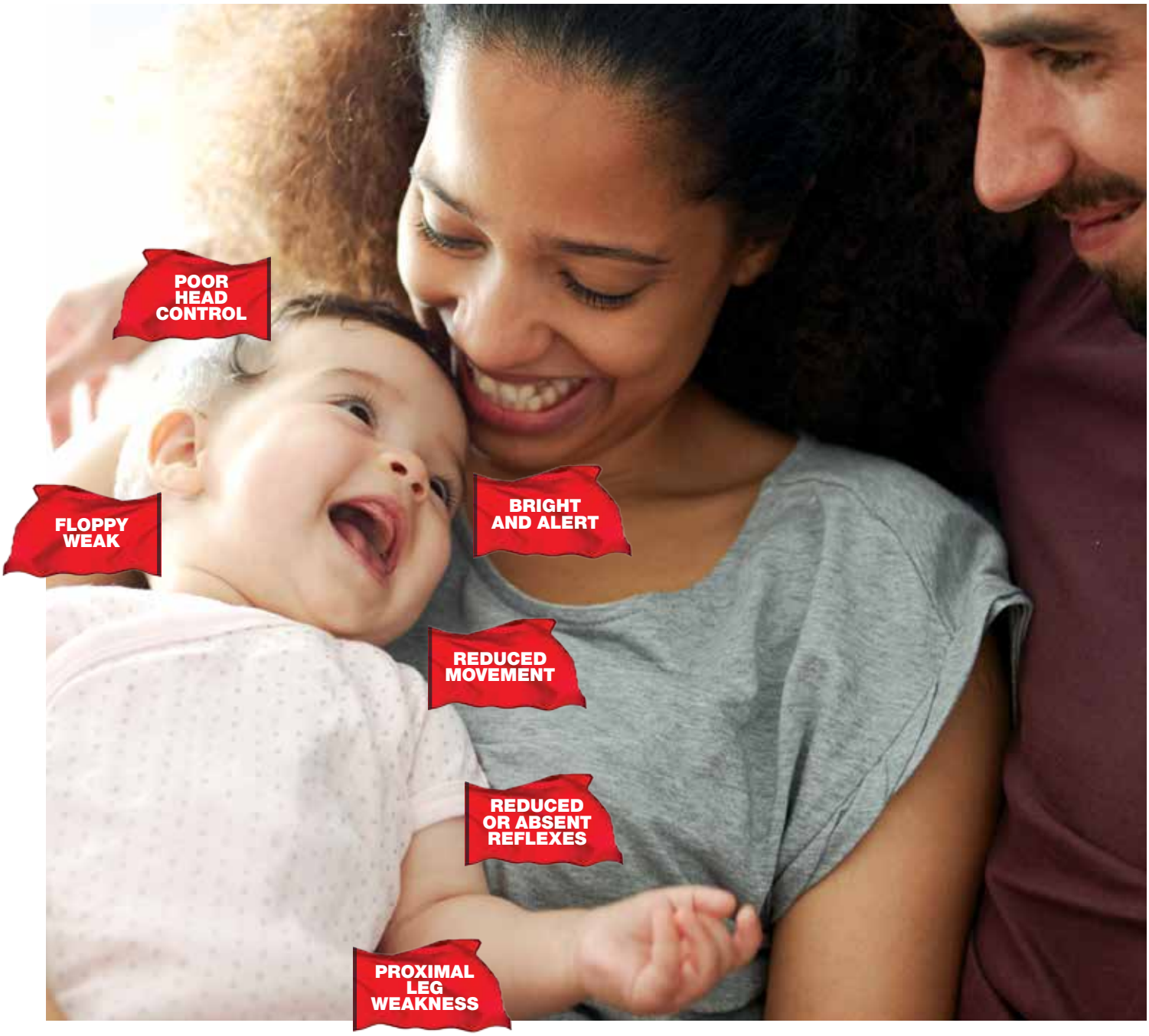
The five domains of general practice

- Communication skills and the patient–doctor relationship
- Applied professional knowledge and skills
- Population health and the context of general practice
- Professional and ethical role
- Organisational and legal dimensions

RACGP



Know the red flags of SMA

Spinal muscular atrophy (SMA) is a rare disease, but early diagnosis is crucial. These are some of the red flags to watch out for in infants.

If you suspect SMA, refer the child today. Specify urgency by querying SMA on the referral form. With new treatments now available for SMA, early diagnosis can make a big difference to survival and function.

For more information, including a CPD activity, visit BewareTheRare.com.au/SMA

RACGP

BEWARE THE RARE

Development of this advertisement was led by the Royal Australian College of General Practitioners, with funding from the Australian Government Department of Health.

About this activity

The Royal Australian College of General Practitioners (RACGP) recognises that general practitioners (GPs) 'play a crucial role in the healthcare and wellbeing of children, young people and their families'.[1]

The most common eye infection, frequently seen in children aged <5 years, is conjunctivitis.[2,3] GPs are well placed to determine whether the cause is viral or bacterial and, therefore, whether the child requires antibiotics.

The gene for spinal muscular atrophy (SMA) is carried by one in 35 Australians, resulting in one out of 10,000 live births being affected.[4] The RACGP recommends that all women planning pregnancy or in their first trimester are offered carrier screening for SMA.[5]

Approximately 2.4% of adolescents aged 11–17 years have reported problem eating behaviours.[6] As it is a newly characterised eating disorder, the incidence of avoidant restrictive food intake disorder is currently unknown. However, it generally commences in infancy or early childhood and can have a significant impact on health if not recognised.[7]

The most common cause of genital discomfort in a prepubertal child is vulvovaginitis. The condition may recur but generally improves with age.[8] GPs can reassure parents that this condition is unlikely to be serious, but can also perform additional testing if there are doubts regarding the diagnosis.

This edition of *check* considers the investigation and management of health concerns in paediatric patients.

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Learning outcomes

At the end of this activity, participants will be able to:

- discuss the investigation of red eye in a child, including possible diagnoses
- identify the red flags that indicate a child may have spinal muscular atrophy
- outline the features that differentiate anorexia nervosa, bulimia nervosa and avoidant restrictive food intake disorder
- discuss the process of diagnosing vulvovaginitis in a child.

Authors

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Abbreviations

ARFID	avoidant restrictive food intake disorder
BMD	bone mineral density
CBT-AR	cognitive behavioural therapy for ARFID
FBE	full blood examination
IVF	in vitro fertilisation
SMA	spinal muscular atrophy
SMN	survival motor neuron
SMN1	survival motor neuron 1
URTI	upper respiratory tract infection

CASE

1 Maddox has red eyes

Maddox is a previously well boy aged three years. He is brought to your practice by his parents, who report he has had worsening red eyes for the past 24 hours.

Question 1

What further history would you like to know about Maddox's red eyes?

Further information

Maddox frequently rubs his eyes during your consultation. His parents tell you they are unsure if he has had any contact with people with similar symptoms. He has felt 'a bit hot' and has a slight runny nose now. When asked to describe what his eyes feel like, Maddox says they feel 'sandy'.

Question 2

What are some red flags and important diagnoses you would want to exclude as you conduct your consultation?

Question 3

How would you examine Maddox and his eyes?

Further information

You are able to test Maddox's visual acuity using the Lea chart, and you are satisfied that he has no visual problems. You cannot see any evidence of a foreign body or corneal abrasion with fluorescein staining. You notice Maddox has watery purulent discharge from both eyes, and his conjunctiva are red bilaterally. You also notice that he has bilaterally raised cervical lymph nodes that are not tender to palpate. The rest of his systems examination is typical.

Question 4

Having ruled out any red flags, what conditions might be causing Maddox's red eyes?

Question 5

How would you differentiate between viral, bacterial and allergic conjunctivitis in order to exercise prudence with antibiotic stewardship?

Further information

You conclude that, as Maddox has had a recent upper respiratory tract infection and currently has enlarged cervical lymph nodes and a watery mucoid discharge from his eyes, he has a viral conjunctivitis.

Question 6

Under what circumstances would you perform any investigations on a child with discharging red eyes?

Question 7

What treatment would you advise for Maddox?

Question 8

What advice would you give to Maddox's parents?

CASE 1 Answers

Answer 1

While it is important to take a full detailed history in paediatrics, a focused ophthalmic history should include the following.

Red flags to exclude:[1]

- headaches
- visual disturbance
- fever
- rash
- trauma to the face
- history of systemic illness.

Further points to consider:

- light sensitivity
- time frame of onset
- contact with people who have similar symptoms
- previous eye problems
- presence of eye pain
- itchiness.

Answer 2

Some of the conditions to include in the differential diagnosis are outlined in Table 1.

Table 1. Differential diagnosis of red eye in a child

Diagnosis	Signs and symptoms
Kawasaki disease	• Conjunctivitis bilaterally – typically non-discharging • Fever • Rash • Cervical lymphadenopathy • Strawberry tongue • Cracked lips • Red and swollen hands and feet[10]
Corneal abrasion	• Pain, watery discharge, photophobia • Conjunctiva may or may not be injected • Usually unilateral but could be bilateral (eg sand grit) • Sudden onset[3]
Corneal foreign body	• Pain, watery discharge, photophobia • Sudden onset • Gritty eye • Usually unilateral[3]
Herpetic dendritic ulcer	• Pain, watering, photophobia • Corneal ulceration without history of trauma • Usually unilateral[1]
Chemical burns	• Sudden onset • Watering • Painful or burning • Child found near chemicals[3]
Iritis	• Dull aching eye pain • Blurred vision • Autoimmune disease[1]
Scleritis and episcleritis	• Painful eye • Blurred vision • Autoimmune disease • Red-purple hue in scleritis[11]
Glaucoma	• Cornea cloudy • Pain, watery discharge, photophobia • Nausea, vomiting, headache • Halos in vision[12]
Trauma	• Subconjunctival haemorrhage, hyphema • Focal redness of conjunctiva • History • Asymmetrical pupils[3]
Orbital cellulitis	• Lid redness • Orbital swelling • Ophthalmoplegia • Proptosis[13]
Measles	• Child unwell with fever and usually miserable • Three Cs: conjunctivitis, cough, coryza • Maculopapular rash (morbilliform) • Koplik spots[14]
Trachoma	• Conjunctival redness, discharge • Irritation of eyes • Eyelid swelling • Inflammation of the inside of the upper eyelid • Enlarged cervical lymph nodes • More common in Aboriginal and Torres Strait Islander populations and in Africa and Asia[15]

Answer 3

Performing an eye examination requires the child's cooperation and practitioner patience. Anesthetising the eye with topical anaesthetic drops may help to calm the child.[2] Most practices will not have a slit lamp; therefore, if detailed and further examination is required, it is recommended to refer the patient to a local emergency department or ophthalmologist who has access to the necessary equipment.

It is always important to remember that a child that is brought to your office may not have seen a doctor for anything other than immunisations. Every encounter with a child is an opportunity for asymptomatic screening of their physical, anthropometric and emotional state.

Paediatric simplified eye examination

A: (Visual) Acuity

Table 2 outlines how to test visual acuity in a child.

Table 2. Tests for examining a child's visual acuity based on age[16–19]

Age	Test
Birth	Aware of light
6–8 weeks	Can fix and follow
6–12 months	Can fix on a toy, reaches in the direction of the object Can use Teller acuity cards
12 months	Can pick up a 1 mm crumb
1–2 years	Can use Cardiff acuity test cards
2–3 years	Can match symbols or shapes on a Lea symbols chart or tumbling E chart
>3 years	Can name letters or pictures on a LogMAR chart or Snellen chart

A: Anaesthesia considered

A: (Surrounding) Anatomy

- Swelling around eyes
- Eyelid laceration and orbit injuries (especially if there is a history of trauma)
- Bruising – exclude globe injury, facial fractures and basal skull fractures
- Orbit fractures – characterised by restriction of extraocular movement

B: Behaviour

- Aversion to bright light
- Rubbing of eyes
- Pain
- Appropriate behaviour for age
- Signs of child abuse

C: Check pupils

- Symmetry of movement of the eyes
- Red reflex
- Size and pupillary reaction
- Visual fields

D: Direct assessment

- Look for foreign body, conjunctival redness and conjunctival haemorrhage

E: Evert

- Evert upper eyelid to exclude subtarsal foreign body

F: Fundoscopy

- Fundoscopy may be difficult to perform without first dilating the pupils or without a slit lamp

F: Fluorescein staining of the eye

- Look for corneal abrasions, ulcerations and foreign bodies

G: Global examination

- Cervical lymph nodes
- Presence of rash
- Full systems examination is recommended – refer to resources available for examination in paediatrics.[2–4]

Answer 4

The most common cause of bilateral red eyes in a child is viral conjunctivitis, bacterial conjunctivitis or allergic conjunctivitis.[1]

Answer 5

Table 3 provides a guide to differentiating between types of conjunctivitis. However, there is considerable overlap in the signs and symptoms displayed.

Children aged 2–12 months with sticky eyes often have blocked lacrimal (tear) ducts rather than conjunctivitis, and the accumulated material is mucus from the tear film rather than pus. If the conjunctiva is not inflamed, this should be treated by frequently removing the mucoid discharge, cleansing the eye with saline and massaging down the path of the nasolacrimal duct. This usually resolves the issue; however, if it has not resolved by 12–18 months of age, a referral to an ophthalmologist for lacrimal duct probing under general anaesthetic may be necessary.[5–7]

Table 3. Differentiating between bacterial, viral and allergic conjunctivitis[20–23]

	Bacterial	Viral	Allergic
Age	Neonates to toddlers	School age to adult	Late childhood to early adulthood
Aetiology	*Staphylococcus aureus, Streptococcus pneumoniae, Haemophilus influenzae* Note: From birth to two weeks of age, consider gonorrhoea and chlamydia – always swab this age group for diagnosis.	Adenovirus Herpes simplex virus – crusting around eyes (note: occurs in immunocompromised patients)	Hypersensitivity reaction to allergen (eg pollen)
Clinical signs	Rapid onset May be unilateral or bilateral Crusting over the eye lashes	Recent viral upper respiratory tract infection (URTI) Pre-auricular lymphadenopathy May initially start with one eye then progress to both eyes Irritated eyes May have household contacts with the same symptoms Psudomembrane visible	Itchy May be a seasonal or chronic condition
Appearance of conjunctiva	Papillary response	Follicular reaction	Cobblestone papillary
Consistency of discharge	Purulent	Watery or mucoid	Watery Sometimes mucoid
Associated disorders	Otitis media	URTI	Eczema, rhinitis, asthma
Contagiousness	Contagious	Very contagious	Not contagious

Answer 6

Usually the diagnosis of a child with discharging red eyes is clinical and does not require swabs. In some circumstances, a swab may be performed if:

- the infection is chronic or recurrent
- herpes simplex virus 1 or 2 is suspected
- the presentation is atypical.

Neonatal conjunctivitis

Red and discharging eyes in a neonate (aged <28 days) have very different likely aetiologies based on age (Table 4). Swabs of the discharging eye are required for microscopy, culture and sensitivity and polymerase chain reaction. It is also imperative to treat the parents if the child is positive for chlamydia and gonococcus. Refer the child to a neonatologist or paediatrician, and notify the Department of Health where relevant.

Table 4. Age-specific aetiologies of conjunctivitis[1]

Age of presentation	Likely aetiology
Day 1	Gonococcal
Day 4–6	Staphylococcal, streptococcal, *Haemophilus* spp.
Day 8–12	Chlamydia
Day 7–14	Herpes simplex virus

Answer 7

There is no cure for viral conjunctivitis. The treatment offered involves symptomatic relieving measures such as:

- cold compress to the eyes
- lubricating preservative-free eye drops
- oral analgesia if the patient is irritable.

There is no role for topical antibiotics for viral conjunctivitis.[8]

Follow-up is recommended after 5–7 days, or earlier if the condition is worsening.

Answer 8

It is recommended to educate parents about the following hygiene measures to reduce the spread of the infection:[9]

- wash your and the child's hands often
- stay home from daycare or school until discharge from eyes has resolved
- avoid touching or rubbing eyes
- frequently clear discharge from around the eyes using a fresh cotton ball for each eye
- discard used moisturisers that may have been in contact with the child's face
- wash face cloths, bed linen and towels in hot water and detergent
- avoid sharing bed linen, towels and clothing
- avoid swimming in a public swimming pool.

Parents can be reassured that the symptoms will resolve on their own with time.

Conclusion

For a case of viral conjunctivitis in a child, thorough history and examination is imperative to make the correct diagnosis to curb the spread of the disease and avoid the unnecessary use of antibiotics.

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CASE

2 Kara seems 'floppy'

Belinda, aged 35 years, has come to see you because her daughter, Kara, aged four months, does not seem to be developing in the same way as her older brother did at that age. Belinda says Kara seems 'floppy'. Kara has trouble holding her head up and lifting her head when lying on her stomach. Belinda is quite worried, as Kara's infant health record indicates she should be doing all these things by this age. Belinda says that 'something is not quite right', and tells you that Kara is not feeding properly and has lost weight. Kara is a bright and interactive infant who smiles appropriately and is interested in her surroundings.

Question 1

Which aspects of Kara's presentation would you be concerned about and why?

Question 2

What conditions would you include in your differential diagnosis?

Further information

You examine Kara, first with her lying on the bed. You notice that she has very little spontaneous kicking of her legs, which are lying in a 'frog leg' position. She has excessive head lag on pull-to-sit. When you pick her up, she has very poor head control, will not take any weight through her legs and tends to slip through your fingers when held under her armpits. She has difficulty holding her head up. In ventral suspension she has an inverted U position.

You notice that Kara has tachypnoea, and when she lies on her back, she displays 'see-saw' breathing (as she breathes in, her chest sucks in and stomach goes out). You check her temperature, and she is afebrile.

Question 3

What would you do next and why?

Further information

You refer Kara to a paediatrician. Because of a number of red flags, you query a neuromuscular disorder, possibly spinal muscular atrophy (SMA), in the referral and request that she is seen quickly, in the next few days.

Question 4

Why is it important to get a prompt referral for Kara?

Further information

The paediatrician requests urgent genetic testing for SMA. A full blood examination is undertaken, and results return in 1–2 weeks. The testing identifies homozygous deletions in the survival motor neuron 1 (*SMN1)* gene, and Kara is diagnosed with SMA type 1.

She begins treatment with nusinersen.

Question 5

What is SMA? What is Kara's prognosis?

Question 6

What other treatments or therapies can you arrange for Kara?

Further information

Belinda asks whether her future children may also develop SMA. She also wonders if Kara's condition could have been identified earlier.

Question 7

What would you tell Belinda?

Further information

Following Kara's diagnosis, Belinda and her partner undergo genetic counselling and carrier testing at the neuromuscular clinic. Both parents are identified as genetic carriers for SMA.

Question 8

What options are available to 'carrier couples' when planning pregnancy?

CASE 2 Answers

Answer 1

Although she is bright, alert and non-dysmorphic, it is concerning that Kara has hypotonia and has not achieved important motor milestones (Table 1).[1] She appears to be 'floppy weak' and might have a neuromuscular disorder.

Answer 2

Muscle weakness is used to distinguish central and peripheral neurological disorders in hypotonic infants ('floppy strong' or 'floppy weak').[2] Assessment of weakness includes the ability to cough and swallow and the quality of spontaneous antigravity movements. Kara's history of having trouble with her head control while being bright and alert raises concern that she is 'floppy weak' and might have a neuromuscular disorder. This is significant even though she is younger than six months of age, as her motor development is different to expectations (siblings and her health record). These conditions affect the peripheral nervous system at varying locations – motor neuron (SMA), peripheral nerve (neuropathy), neuromuscular junction or muscle fibres (myopathy or dystrophy) – and result in muscle weakness.

While the severity of signs and symptoms varies between individuals, the common theme for paediatric neuromuscular disorders is the presence of muscle weakness and impaired motor function.[3] Although paediatric neuromuscular disorders are rare, the more common conditions are:[4]

- SMA – approximately one in 10,000 live births
- Duchenne muscular dystrophy – approximately one in 5000 boys
- Charcot-Marie-Tooth disease – approximately 15.7 per 100,000 births.

Table 1. Gross motor milestone red flags by age[1]*

Six months	Nine months	12 months	18 months	Two years
Not holding head and shoulders up with good control when lying on stomach Not holding head with control in supported sitting	Not rolling Not sitting independently/without support Not moving (eg creeping, crawling) Not taking weight on legs when held in standing position	No form of independent mobility (eg crawling, commando crawling, bottom shuffle) Not pulling to stand independently and holding on for support	Not standing independently Not attempting to walk without support	Not able to walk independently Not able to walk up and down stairs holding on

*Aid suitable for children who were born on or after 38 weeks' gestation. If born prior, use adjusted age for milestones. Table adapted from Child Development Program and Brisbane North Primary Health Network, Red flags early identification guide, Brisbane, Qld: Child Development Program and Brisbane North Primary Health Network, 2016, available at www.childrens.health.qld.gov.au/wp-content/uploads/PDF/red-flags-a3.pdf [Accessed 4 September 2020].

The differential diagnosis is a 'floppy strong' infant, in whom antigravity movement is present. This may be due to a central nervous system disorder (eg Down syndrome). The infant may have other abnormal brain functions; for example, decreased consciousness or seizures.

Answer 3

Kara is 'floppy weak' and appears to have some of the characteristic red flags for SMA.

The red flags for SMA include:

- hypotonia (relaxed tone)
- poor head control (eg head lag when pulled to sit, if aged >2 months)
- 'frog leg' posture when lying – not lifting legs at the hips to kick
- tongue fasciculations
- reduced or absent deep tendon reflexes
- muscle weakness
- delayed motor milestones (Table 1)
- bright and alert.

It is important to refer her urgently to a paediatrician or, preferably, a paediatric neurologist for assessment.

Answer 4

The 'wait and watch' approach to delays in motor development is no longer appropriate in light of new treatments available. For conditions such as SMA, it is important to prevent delays in diagnosis, allowing for timely intervention and treatment. A new medication, nusinersen, is available to treat the condition – and the earlier treatment is initiated, the better the outcome.[5]

Nusinersen is a synthetic antisense oligonucleotide that promotes increased production of functional survival motor neuron (SMN) protein and has changed the treatment landscape for SMA.[5] Nusinersen was added to the Pharmaceutical Benefits Scheme in 2018 for the treatment of infantile or childhood-onset SMA. It is administered via intrathecal injection (12 mg nusinersen/dose) at a tertiary neuromuscular centre, using a 22-gauge or 24-gauge needle at 1, 15, 30 and 60 days, followed by ongoing four-monthly maintenance doses.[6] Children undergoing treatment may show improved motor function and survival.[5]

Answer 5

SMA is a neuromuscular disorder that ranges in severity from progressive infantile paralysis and premature death to full life expectancy. It is caused by a mutation in the *SMN1* gene, leading to a deficiency of SMN protein. Subsequent loss of motor neurons in the brainstem and spinal cord lead to muscle denervation and atrophy, physical disability and respiratory insufficiency.[7]

Patients with SMA are grouped according to age of onset and maximal motor milestones. SMA type 1 (onset 0–6 months) accounts for 50–60% of cases. SMA types 2 (onset 6–18 months) and 3 (onset >18 months) represent 30–40% of cases.[8]

Without treatment, patients with SMA type 1 tend to experience rapid disease progression and respiratory complications such as aspiration pneumonia; life expectancy is <2 years.

However, if treated with nusinersen, children may show improved motor function and survival.[5] Early treatment is recommended – the greatest benefits are seen in children treated in the pre-symptomatic phase, emphasising the importance of early diagnosis.[9]

Answer 6

Because she has significant weakness at diagnosis, Kara will likely experience ongoing weakness and mobility issues. It is important that multidisciplinary supportive care is also provided, including early and active maintenance of motor function through physiotherapy and physical assistance, respiratory management and nutritional optimisation.[10,11] A tertiary paediatric neuromuscular clinic typically includes a neurologist, respiratory physician, geneticist, physiotherapist, occupational therapist, dietitian and speech pathologist to implement best practice care. The goal is to optimise function and anticipate and mitigate complications.

As a general practitioner (GP), your role is to provide guidance for management of acute illnesses at home,

advocate for support with the National Disability Insurance Scheme, provide support to the family and assist with routine vaccinations. Annual influenza immunisation should also be administered. Kara is particularly vulnerable to deterioration with community-acquired infections and will require careful assessment when unwell. Her paediatric neuromuscular team will provide an individualised anticipatory care plan for assessment and management of acute illnesses at home. This will include vital signs and symptoms that will guide an intensification of care, with recommendations for airway clearance, ventilation, antibiotics, hydration and emergency contacts.

Answer 7

SMA is not identified by 'routine' pregnancy care, for example by the non-invasive prenatal test, triple test, ultrasonography or newborn screening (heel prick).

SMA is an autosomal recessive disease, and the recurrence risk is 25%. Genetic carrier screening for SMA can be offered to couples who have previously had an affected child. A GP or a genetics counsellor associated with the tertiary neuromuscular clinic can provide education, testing and follow-up to facilitate future informed reproductive decision making.

Further resources for GPs include The Royal Australian College of General Practitioners' Continuing Professional Development modules on the Beware the Rare website (https://bewaretherare.com.au).

Answer 8

The options available to 'carrier couples' include:[12,13]

- proceeding to a pregnancy, accepting the possibility that their child may be affected – in which case, they may be better placed to optimise the child's health and wellbeing (through early diagnosis and intervention)
- undergoing fetal diagnostic testing (via amniocentesis or chorionic villus sampling) and terminating affected pregnancies or preparing for the possibility that they may give birth to an affected child
- undergoing in vitro fertilisation (IVF) with pre-implantation genetic testing to screen embryos before they are implanted in the uterus
- undergoing IVF with donor eggs, sperm or embryos
- choosing to adopt or not have children.

The majority of children with SMA are born to couples with no previously known family history. Therefore, offering genetic carrier screening to only adults with a family history of SMA will only identify a minority of carrier couples. The Royal Australian and New Zealand College of Obstetricians and Gynaecologists now recommends that all women or couples planning a pregnancy, or in the first trimester of pregnancy, should be offered information about the availability of carrier screening for the more common genetic conditions, including SMA.[12]

Resources for doctors and patients

- Beware the Rare – Information for health professionals and patients, including Continuing Professional Development activities, https://bewaretherare.com.au
- SMA Australia, smaaustralia.org.au

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CASE

3 Tilly is underweight

Tilly, aged 13 years, is brought to your clinic by her mother, Jane. Jane has been concerned about Tilly's eating and her lack of weight gain for some time; however, this has become more obvious since Tilly started secondary school and her friends now 'tower over her'. She has also been teased for looking like 'a primary school kid'. Tilly's younger brother, aged 11 years, is the same height as she is.

Question 1

What further history would be useful to attain from Jane?

Further information

Tilly was born at 36 weeks' gestation and weighed 2.8 kg. She has not had any serious illnesses, and she achieved her developmental milestones within the usual timeframes. She was breastfed until 12 months of age, then weaned onto solids without any difficulty. Jane reports that Tilly was always a small child. While Tilly has always been interested in food, she has typically eaten small amounts. This was most noticeable when she started kindergarten. She has never been able to finish her food; when asked to eat more, Tilly would complain of feeling so full that she had abdominal pain and was afraid that she may vomit. Jane has also noticed that Tilly has been eating less of her lunch at school. Tilly has yet to start menstruating.

Question 2

What history would you like to obtain from Tilly?

Further information

Tilly herself reports that she does get hungry but simply gets full quickly. She does not complain of abdominal pain at other times. The abdominal pain is not related to any particular food and only occurs at the end of the meal when she is full. She does not have reflux and has not vomited. She has no other physical symptoms, in particular constipation or diarrhoea.

Tilly worries that she looks smaller and younger when compared with her peers. She has not been dieting. In fact, she would like to gain weight in order to grow. She does not exercise, preferring to do art and craft instead. Tilly tells you that she tends to worry about assignments and schoolwork. Jane reports that Tilly is perfectionistic and achieves good marks.

Tilly further reports that the transition to secondary school had been difficult as she was the only one from her primary school who moved to the new school. Being shy, it took her a long time to make friends. She also worries about what her peers think about her and admits to overthinking about what friends or classmates have said. Sometimes she feels sad when her friends do not get along with each other.

On examination, Tilly is a small, thin, neatly dressed, shy teenager who appears younger than her age. She has pressured speech. The growth chart shows that Tilly's height is 148 cm and has always been on the tenth centile. Her weight is 28.8 kg and has been on the third centile until the past 12 months; it is now below the first centile. Her body mass index is 13.1 kg/m^2 (below the first centile). Tilly's pulse rate is 90 beats per minute and her blood pressure is 90/55 mmHg with no postural changes. She has a temperature of 36.8°C. Her abdomen is soft and non-tender with no masses. The rest of the physical examination does not reveal any abnormalities.

Question 3

What investigations might you perform?

Further information

Tilly has a full blood examination (FBE), liver function tests and testing for electrolytes, calcium, magnesium and phosphate, all of which are normal. She has a borderline low ferritin level but normal levels of vitamins B12 and D, folate and zinc. She also has a normal blood sugar level and urinalysis, erythrocyte sedimentation rate, coeliac screen and thyroid function tests.

Her oestradiol is <50 pmol/L (reference range: 0–936 pmol/L), but her follicle-stimulating hormone and luteinising hormone levels are within normal limits.

Tilly has a delayed bone age, consistent with the age of 11 years and six months. Her bone mineral density (BMD) is also low, with a height-adjusted lumbar BMD z-score of –2.6, a right hip BMD z-score of –2.8 and a total BMD (minus head) z-score of –2.3.

Question 4

What conditions would you consider in your differential diagnosis?

Question 5

Could Tilly just be anxious? What mental health comorbidities may be associated with eating disorders such as avoidant restrictive food intake disorder (ARFID)?

Question 6

What would be your approach to managing Tilly?

CASE 3 Answers

Answer 1

Aside from a full medical history that includes Jane's pregnancy and Tilly's birth history, past medical problems, family history, immunisation, allergies and current medications, a targeted history focusing on Tilly's eating, growth and development is useful.

A longitudinal history of Tilly's eating habits will help determine how persistent the problem has been. It is also helpful to determine the range or variety of accepted foods across the basic food groups and the adequacy of energy intake.

A 24-hour dietary review can aid this discussion. For some children and young people it is also helpful to enquire about the appearance, smell, colour or texture of food that is being avoided. The degree of distress or functional impairment at mealtimes in the family or other social settings can provide additional information.

You can also ask Jane about Tilly's growth over the past 13 years, enquiring about whether there have been significant changes such as weight stagnation or loss, and if this could be linked to any particular events. This can be reviewed together with the growth chart.

Answer 2

It is important to obtain a history from the child or young person to ascertain their own perspectives about their eating. Querying attitudes towards weight and shape, the drive to exercise and any compensatory behaviors (such as purging or laxative use) helps with excluding other eating disorders such as anorexia nervosa and bulimia nervosa. The SCOFF screening questionnaire may be helpful in doing so.[1] Taking a psychosocial history using the HEEADSSS (Home environment, Education and employment, Eating, peer-related Activities, Drugs, Sexuality, Suicide/depression, and Safety from injury and violence)[2] framework allows Tilly's eating difficulties to be placed in the broader context of her life at home, school and friendships as well as providing information about any underlying mood disturbance.

Answer 3

It is recommended that most patients undergo an FBE; testing of electrolytes including calcium, magnesium and phosphate; liver function tests; erythrocyte sedimentation rate testing; measurement of blood sugar level as well as urinalysis. It is worth screening for other causes of weight loss such as coeliac disease and thyroid dysfunction. Depending on the diet history, screening for micronutrient deficiency such as iron, vitamins B12 and D, folate and zinc can also be undertaken. Follicle-stimulating hormone, luteinising hormone and oestradiol can be checked in females who present with amenorrhoea.

Stool microscopy and calprotectin can be considered if there are concerns about inflammatory bowel disease**.** Abdominal pain associated with diarrhoea (often with mucus and blood),

fatigue and weight loss may be suggestive of inflammatory bowel disease. Barium swallow/meal or endoscopy may also be a consideration depending on the history and presentation. Note, however, that patients with ARFID and other eating disorders commonly complain of somatic symptoms such as dysphagia or bloating, fullness and abdominal pain related to delayed gastric emptying and slow transit constipation, especially if they are malnourished. Depending again on presenting symptoms and signs, more extensive investigations may be performed to exclude other causes of weight loss, for example endocrine conditions (type 1 diabetes, Addison's disease), malignancies and chronic infections. It is important to note that ARFID can co-exist with chronic conditions.

Electrocardiography should be performed to look for electrophysiological abnormalities associated with weight loss (small complex, QTc) or vomiting associated with hypokalemia (U waves).

Longstanding undernutrition can affect growth and bone health, hence the assessment of bone age and bone density (dual-energy X-ray) at baseline and at annual/biannual intervals can help evaluate the consequences.[3]

Answer 4

Tilly's longstanding history of eating only small amounts and the lack of body image distortion or drive for thinness would exclude anorexia nervosa. There is also no history of overvaluation of shape, binge eating or compensatory behaviours suggestive of bulimia nervosa. Tilly's symptoms and history suggest she has ARFID.[4] Table 1 summarises the differences between these conditions.

Table 1. Differences between anorexia nervosa, bulimia nervosa and avoidant restrictive food intake disorder

	Anorexia nervosa/atypical anorexia nervosa	Bulimia nervosa	Avoidant restrictive food intake disorder
Significant weight loss/failure to gain weight	Yes	No	May or may not be present
Underweight	Yes/Not yet	No	May or may not be present
Body image disturbance	Yes	Yes	No
Binge eating	Maybe	Yes	No
Overexercising/ laxatives/fasting/ vomiting	Maybe	Yes	No

ARFID emerged as a new diagnostic category in the *Diagnostic and Statistical Manual of Mental Disorders,* fifth edition (DSM-V)[4] with the reformulation of the DSM-IV category 'Feeding disorders of infancy and early childhood'.[5] Despite this, patients with feeding and eating concerns across the life spectrum have long been recognised in the primary care setting. The diagnostic criteria for ARFID are outlined in Table 2.

Table 2. Diagnostic criteria for avoidant restrictive food intake disorder (ARFID)[6]

What ARFID is	What ARFID is not
• A persistent problem with feeding or eating leading to an inability to take in adequate nutrition/ coupled with one of the following: – substantial weight loss or failure to gain weight in growing children/adolescents – major nutritional deficiency – dependence on oral nutritional supplement or nasogastric tube feeds – marked interference with psychosocial functioning	• Associated with body image disturbance • Due to scarcity of food • Related to culturally sanctioned practice • Explained by another medical problem or psychiatric disorder, such that the eating problem resolves with the treatment of the medical or psychiatric disorder

ARFID is a complex and heterogenous disorder that can present with multiple physical symptoms and comorbid medical conditions and psychiatric problems.[6] Common clinical presentations of ARFID include sensory sensitivity, lack of interest and fear of aversive consequences.[7]

Sensory sensitivity

Some patients report a heightened sensitivity to food (appearance, taste, texture, smell or colour) and hence avoid these foods, often eating from a narrow range of selected foods. Some patients in this group avoid trying new foods (food neophobia).

Lack of interest

Patients in this group report a low interest in food and/or a poor appetite. They often describe not feeling hungry at meal times, forgetting to eat and/or feeling full more quickly than others. Often the diet is low in volume, which maintains the disorder.

Fear of aversive consequences

Patients in this category avoid foods because of the fear of choking or vomiting, abdominal pain or perceived harmful effect of food, for example contamination. Often, food-related trauma has been experienced in the past, resulting in subsequent avoidance of the index food. This may then further generalise to other foods. Ongoing restriction and further weight loss then affects gastric emptying, which subsequently makes eating regularly more challenging. This can present acutely with rapid weight loss.

It is also common for patients with different clinical presentations of ARFID to have overlapping characteristics. When compared with patients who have anorexia nervosa and bulimia nervosa, clinical studies show that patients with ARFID are more likely to be male and tend to present at a younger age. They are also more likely to have a longer history prior to presentation.[5]

Answer 5

It is important to assess Tilly for a comorbid anxiety disorder, which is commonly seen in patients with ARFID. It would appear that transition to secondary school and peer relationships have exacerbated Tilly's anxiety. A retrospective chart review of 34 paediatric patients with ARFID indicated that 50% also had generalised anxiety disorder.[8] Other related psychiatric comorbidities include obsessive compulsive disorder, mood disorders, autism spectrum disorder and attention deficit hyperactivity disorder.[6]

A referral to a psychologist or child and adolescent psychiatrist with experience or interest in eating disorders or an eating disorder service may help confirm the diagnosis and comorbid disorder(s).

Answer 6

As ARFID is a relatively new diagnosis, evidence-based strategies are currently being trialled, and consensus guidelines for treatment have yet to be fully developed. As with other eating disorders, a multidisciplinary approach is helpful. The treating team can comprise a medical practitioner, mental health specialist and/or a dietitian skilled in eating disorders. Increasingly, many specialist eating disorder units are also treating patients with ARFID.

Many patients, such as Tilly, have had longstanding low weight such that they have developed compensatory mechanisms and tend not to present with haemodynamic instability. Such patients can be managed in an outpatient setting. Patients who experience acute weight loss may require admission to hospital if they become haemodynamically compromised.

Psychoeducation for both Tilly and her parents is important for them to understand ARFID and treatment approaches. With the help of a dietitian and/or psychologist, Tilly's parents can be guided to gradually increase Tilly's food intake while helping to manage her anxiety about abdominal discomfort and fear of vomiting. Providing regular, scheduled meals and snacks while increasing the caloric density of food to maximise each bite and/or the use of oral supplements can also assist with weight gain. Cognitive behavioural therapy may be useful in helping Tilly with her anxiety.[9]

Tailoring approaches may be required; for example, trying to increase intake using preferred foods initially for those in the sensory sensitivity group before gradually expanding the range of foods, together with the use of behavioural rewards and anxiety management.[10] For those who are avoiding foods because of aversive consequences, systematic desensitisation using graded exposures alongside cognitive therapy to address anxiety simultaneously has been shown to be helpful in restoring nutrition.[11] Ongoing longitudinal medical monitoring is necessary to ensure nutritional rehabilitation and weight gain, resumption of growth and puberty.

There are a number of promising structured approaches tailored specifically for the treatment of ARFID. These include family-based treatment (used primarily for patients with anorexia nervosa and atypical anorexia nervosa)[12,13] and cognitive behavioural therapy for ARFID (CBT-AR),[14] which are currently being implemented and evaluated.

According to guidelines from the Royal Children's Hospital Clinical Practice Guidelines in conjunction with the Victorian Paediatric Clinical Network, admission should be considered for children and young people if the following are present:[15]

- significant electrolyte disturbance (K <3.0 mmol/L)
- heart rate ≤50 beats per minute (bpm)
- postural heart rate increase ≥30 bpm
- resting systolic blood pressure ≤80 mmHg
- postural systolic drop ≥20 mmHg
- hypothermia <35.5°C
- dehydration
- arrhythmia or prolonged QTc >0.45 s
- weight <75% of their expected body weight or rapid weight loss (>10–15% in 3–6 months is significant)
- out-of-control eating disorder compensatory behaviours (eg prolonged fasting/inability to eat at home/uncontrolled purging and exercising).

Admission may be appropriate in rare circumstances where community management is not effective.

Conclusion

Although ARFID is a new DSM-V diagnosis, patients have been long recognised in primary care to experience significant difficulties in eating without body image concerns. While patients may not present acutely with haemodynamic instability, there can be significant medical consequences of longstanding malnutrition. There are now promising approaches to treatment; however, further research is required.

Resources for parents

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- Eating Disorders Victoria, www.eatingdisorders.org.au
- Eating Disorders Families Australia, www.edfa.org.au

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CASE

4 Sarah has genital symptoms

Sarah, aged three years, is brought to see you by her mother, Fiona, who is distressed because Sarah returned from her father's house last night and promptly complained of a sore vagina. Fiona is worried that Sarah was sexually assaulted by her father.

Question 1

How would you approach this consultation?

Further information

You establish that Sarah is a developmentally normal well child. She started toilet training six months ago and is dry in the day but still wears a nappy at night. Sarah is a fiercely independent child who likes to take herself to the toilet. She sometimes forgets to wipe after going to the toilet. Prior to toilet training, Sarah was prone to terrible nappy rash. However, this has resolved since she has been toilet trained.

For the past two days, Sarah has been complaining of burning when she passes urine, and Fiona has noticed a small amount of white discharge on Sarah's underwear. Sarah has complained of burning with urination once or twice before but never this frequently.

Sarah's parents separated after an episode of domestic violence during which both parents were physically violent towards each other. There were never concerns about Sarah's father's behaviour towards Sarah, and Sarah spends alternate weeks with each parent.

Question 2

How would you examine Sarah (including her genitals)?

Further information

Sarah has normal anogenital examination findings, and there is no sign of genital injury. You notice that Sarah's vaginal vestibule and the medial aspects of the labia look quite red. There is no offensive discharge.

Question 3

What is the most likely diagnosis?

Question 4

What investigations do you need to perform?

Question 5

What are the symptoms of vulvovaginitis?

Question 6

What are the causes of vulvovaginitis?

Question 7

How would you treat Sarah's vulvovaginitis?

Question 8

How should you answer Fiona's question regarding whether Sarah has been sexually assaulted by her father?

Further information

Four weeks later, Sarah is once again brought into your clinic. Fiona is very pleased that Sarah's vaginal discomfort disappeared after a couple of vinegar baths, but she has now noticed two blisters next to Sarah's vagina. The blisters do not seem to bother Sarah.

You know from Sarah's last appointment that her parents both have a history of cold sores. Sarah's mother has not previously had a sexually transmissible infection, but you do not have information about Sarah's father.

You decide to swab the lesion, which comes back positive for herpes simplex virus type 1.

Question 9

Is this evidence that Sarah has been sexually assaulted?

CASE 4 Answers

Answer 1

It is recommended to take a comprehensive history including:

- history of presenting complaint – including details about the vaginal pain and the presence of associated symptoms such as itch, vaginal discharge, redness around the vagina and burning or stinging when passing urine
- past history – including previous episodes of vagina pain, previous threadworm infections and known skin conditions such as eczema
- immunisations and medications
- family history
- social history – explore Sarah's relationship with her father, and Sarah's parents' current relationship. Document any court-assigned parenting orders.

Answer 2

Sarah should have a complete examination, from top to toe.

Examining Sarah's top half and lower half sequentially, so that she is partially clothed throughout the whole examination, will help to keep Sarah more comfortable.

Genital examination

At three years of age, and being an independent child, Sarah might be quite uncomfortable about having you look at her genitals. Reassurance from her mother and distraction will be helpful.

The ideal position to look at Sarah's vagina will be on her back with her legs making butterfly wings. Use gentle traction on the labia majora to expose the clitoral hood, vaginal vestibule, hymen and urethra. To complete the examination, flex Sarah's legs up onto her chest to look at her anal region.

Answer 3

The most likely diagnosis is vulvovaginitis, which is the inflammation of the vagina and vulva. It is a common

problem in prepubertal children. The lining of the vagina and vulva is quite thin in young children, and it can be easily irritated. Most cases of vulvovaginitis are of non-specific aetiology. Vulvovaginitis can occur multiple times in the same child.[1–3]

Answer 4

As Sarah is a well child who does not have a fever, no investigations are necessary at this point. This is a clinical diagnosis. When patients with vulvovaginitis complain of dysuria, a urine sample might be indicated. As Sarah is well, this is not urgent.

Answer 5

Symptoms of vulvovaginitis include itching, discharge, redness between the labia majora and burning or stinging with passing urine.[1,2]

Answer 6

Anything that causes moisture around the vulva can cause vulvovaginitis. In most cases the cause is non-specific.

Exacerbating factors include:[1]

- tight clothing, including tight underwear
- increased weight
- chemical irritants – soap, bubble baths, antiseptics.

Threadworms can cause vulvovaginitis and should be considered in all patients, especially those with persistent symptoms and nocturnal itching.[1,3,4]

Bacterial infections, vaginal foreign bodies and chronic skin conditions are less common causes of vulvovaginitis.[4]

Answer 7

Management should follow a conservative approach such as avoiding irritants and exacerbating factors. Vinegar baths provide symptomatic relief for many patients and can be had daily until symptoms resolve. Worm treatments from the pharmacy might be considered (eg pyrantel, mebendazole), particularly if symptoms are nocturnal and not resolving.[1,2,4,5]

Answer 8

It is recommended to explain to Fiona that you cannot say that Sarah has not been sexually assaulted, but that Sarah's genitals look normal and that there is no sign of injury. Advise Fiona that children who have experienced sexual assault as a young child might talk about it when they are ready.

Fiona should be advised that redness around the vagina is common for girls in Sarah's age group and that it is most likely due to vulvovaginitis. Providing Fiona with information about the condition (such as the Royal Children's Hospital fact sheet about vulvovaginitis, www.rch.org.au/kidsinfo/fact_sheets/Vulvovaginitis) and reviewing this information with her can also be helpful.

It is also important to discuss with Fiona that if she continues to have concerns that Sarah has been sexually assaulted by her father for reasons other than her reddened vagina, then she should notify Child Protection (https://services.dhhs.vic.gov.au/child-protection-contacts).

A follow-up appointment for Sarah would be beneficial to ensure that her symptoms are resolving and to help support her mother.

Answer 9

No. While sexual assault needs to be considered, it is important to recognise that children, especially those still wearing nappies, can develop herpetic lesions on their genitals. Transmission can occur from parents or caregivers such as childcare workers performing normal cares.[6] All children with genital herpes should be considered for discussion with the local paediatric forensic medical service.

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ACTIVITY ID **224718**

Paediatric health

This unit of *check* is approved for six CPD Activity points in the RACGP CPD Program. The expected time to complete this activity is three hours and consists of:

- reading and completing the questions for each case study
  - you can do this on hard copy or by logging on to the RACGP website (www.racgp.org.au), clicking on the My Account button and selecting the *gplearning 2020* link from the drop-down
- answering the following multiple choice questions (MCQs) by logging on to the RACGP website (www.racgp.org.au), clicking on the My Account button and selecting the *gplearning 2020* link from the drop-down
  - you must score ≥80% before you can mark the activity as 'Complete'
- completing the online evaluation form.

You can only qualify for CPD points by completing the MCQs online; we cannot process hard copy answers.

If you have any technical issues accessing this activity online, please contact the *gplearning* helpdesk on 1800 284 789.

If you are not an RACGP member and would like to access the *check* program, please contact the *gplearning* helpdesk on 1800 284 789 to purchase access to the program.

Case 1 – Jiang

Wei Hua has come to see you as her son Jiang, aged four months, 'does not seem to be hitting milestones when his sister did or when other kids his age do – he is floppy and something is not quite right'.

Question 1

Which one of the following is a red flag that suggests a 'floppy weak' child, who may have a neuromuscular disorder?

A. Muscle stiffness

B. Hypertonia

C. Reduced alertness and response

D. Not lifting legs at the hips to kick/'frog legs'

Further information

As part of your differential diagnosis, you consider your knowledge of spinal muscular atrophy (SMA).

Question 2

Why is early referral and diagnosis of SMA important?

A. To confirm the diagnosis urgently with genetic testing

B. To start early intervention and multidisciplinary care

C. To facilitate genetic counselling for the family

D. All of the above

Case 2 – Eleni

Eleni, aged two years, is brought to see you by her mother, Anita, who says that Eleni has been crying when passing urine. Anita has noticed that Eleni's vulva looks red and irritated when she has changed her nappies but thinks this is different to the nappy rash that Eleni had as a young baby. You undertake a further history and physical examination and consider vulvovaginitis as part of your differential diagnosis.

Question 3

Which one of the following investigations would you perform in a child with mild vulvovaginitis?

A. No investigations are required

B. Urine culture

C. Bacterial swab

D. Viral swab

Question 4

Which one of the following treatments should be considered for a child with persistent vulvovaginitis and nocturnal itching?

A. Antibiotics

B. Worm treatment (eg pyrantel, mebendazole)

C. Steroid creams

D. Persistence with conservative measures only

Further information

Several weeks later, Anita returns with Eleni and is quite concerned as Eleni has developed a small blister next to her vagina. Investigations confirm the lesion is positive for herpes simplex virus type 1. You consider what action you should take.

Question 5

Which one of the following is the most reasonable approach regarding a child of nappy-wearing age who has a herpetic genital lesion?

A. Contact child protection services immediately

B. Take a further history about the child's caregivers and consider discussing the child with the local forensic service

C. Advise the child's parents that their child has been sexually assaulted

D. No further action is required

Case 3 – Caden

Caden, aged seven years, comes to your general practice with his father. Caden has a history of red eyes, and on examination he tells you both his eyes are really sore.

Question 6

Which one of the following symptoms would prompt immediate referral to an ophthalmologist?

A. Symptoms not resolving with chloramphenicol

B. Bilateral red eyes

C. Severe pain in eyes

D. Difficulty keeping eyes open

Case 4 – Poppy

Poppy was born at term in a regional hospital. Her mother had limited antenatal care through her pregnancy. Poppy had an uncomplicated neonatal course, but it was noted on day nine that she had a purulent discharge from both of her eyes.

Question 7

Which one of the following is the most likely the cause of the discharge?

A. Gonococcus

B. Chlamydia

C. Herpes simplex virus

D. *Staphylococcus aureus*

Case 5 – Jameson

It is late spring and Jameson, aged eight years, presents to your practice with a three-week history of red and watery eyes that feel itchy and gritty. You have seen him once before for a review of his eczema.

Question 8

Which one of the following is the most likely diagnosis?

A. Bacterial conjunctivitis

B. Viral conjunctivitis

C. Allergic conjunctivitis

D. Bilateral foreign body in the eyes

Case 6 – Nawal

Nawal is a young medical student who is attending your practice. She is interested in a career in paediatric health, specialising in eating disorders. She wishes to learn more about the differences between anorexia nervosa, bulimia nervosa and avoidant restrictive food intake disorder (ARFID).

Question 9

Which one of the following factors is a possible presenting symptom for a patient with ARFID?

A. Binge eating

B. Body image disturbance

C. Significant weight loss/failure to gain weight

D. Fasting and/or vomiting

Question 10

When compared with patients who have anorexia nervosa and bulimia nervosa, patients with ARFID are more likely to be:

A. female and tend to present at an older age

B. female and tend to present at a younger age

C. male and tend to present at an older age

D. male and tend to present at a younger age.

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