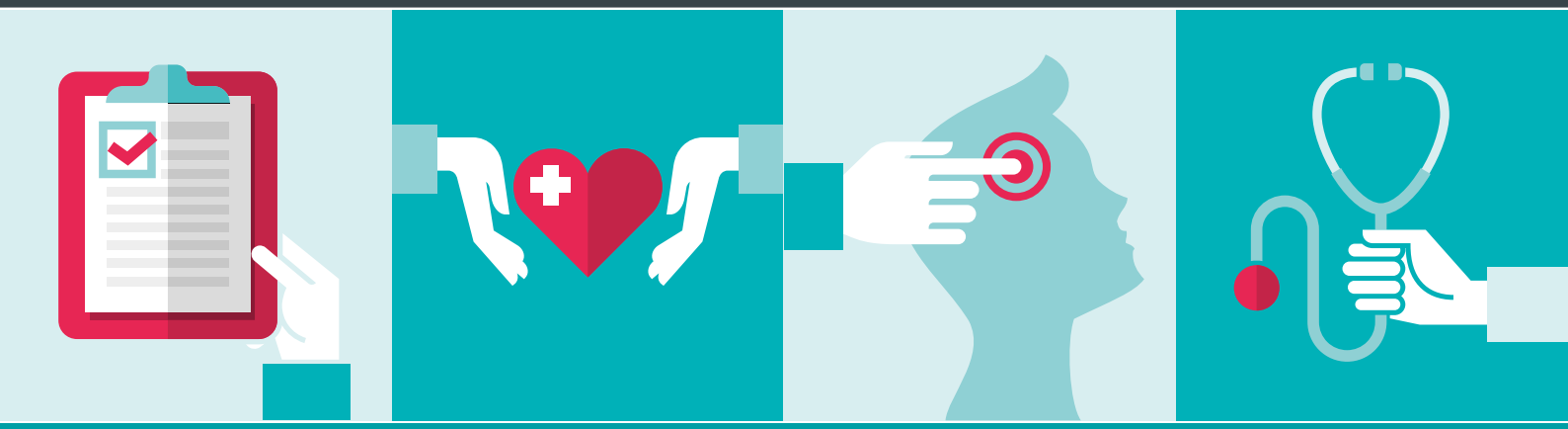
check

Independent learning program for GPs



Unit 515 April 2015

Stages of life: Childhood

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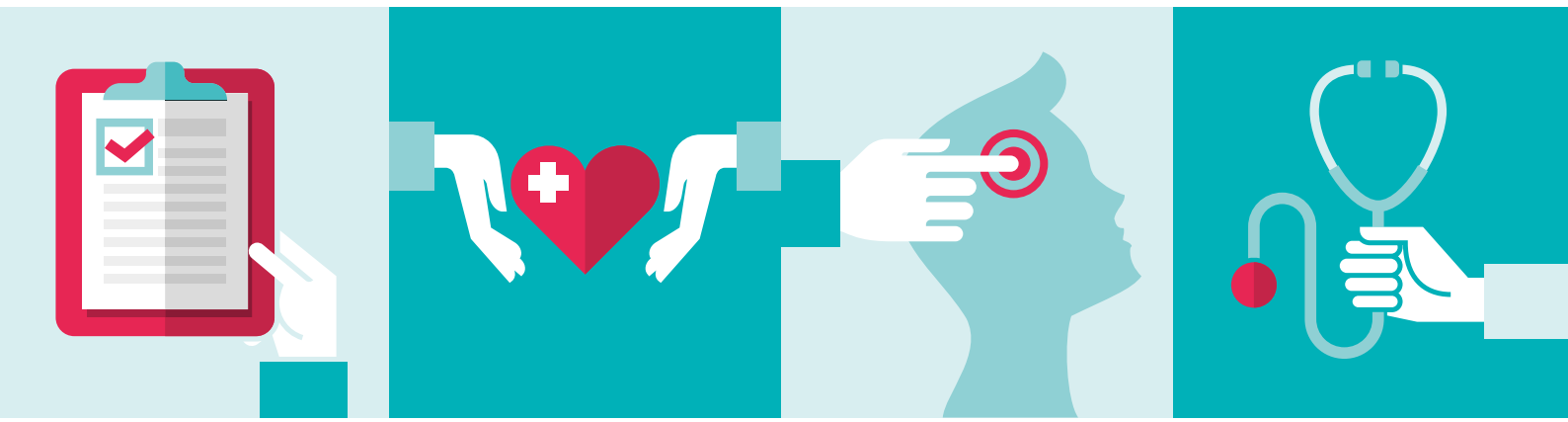

Stages of life: Childhood

Unit 515 April 2015

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

Child mortality rates in Australia have decreased by more than half in the past 20 years.[1] However, chronic illness continues to be a major contributor to the burden of disease in children and about 2 in 5 children in Australia have a long-term condition.[2] Asthma, cancer and diabetes are three chronic conditions of particular concern and are National Health Priority Areas.[2] Preventive measures, early detection and intervention in the areas of health, safety and education can improve a child's health and general wellbeing. This edition of *check* focuses on the assessment and management of childhood conditions in general practice.

LEARNING OUTCOMES

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

- explain how asthma is diagnosed and managed in children
- outline the diagnosis and management of coeliac disease in children
- describe the assessment of children presenting with abdominal pain
- discuss the causes of reflux in infants
- discuss appropriate measures in the assessment of bruising in children
- list the referral pathways for children with learning difficulties.

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ACRONYMS

ADHD	attention deficit hyperactivity disorder
ASRS	Autism Spectrum Rating Scales
CMPA	cow's milk protein allergy
CRP	C-reactive protein
DGP	deamidated gliadin peptide
DSM-5	Diagnostic and Statistical Manual of Mental Disorders, fifth edition
FBE	full blood evaluation
GOR	gastro-oesophageal reflux
GORD	gastro-oesophageal reflux disorder
HLA	human leukocyte antigen
IBD	inflammatory bowel disease
IBS	irritable bowel syndrome
NICE	National Institute for Health and Care Excellence
PBS	Pharmaceutical Benefits Scheme
PEDS	parents' evaluation of developmental status
PPI	proton pump inhibitor
PTSD	post-traumatic stress disorder
SLD	specific learning disorder
tTG	tissue transglutaminase antibody
T1DM	type 1 diabetes mellitus
URTI	upper respiratory tract infection

CASE 1

DANIEL HAS A COUGH AND SHORTNESS OF BREATH

Maria, a lawyer aged 34 years, has made an appointment to see you to discuss her son Daniel who is 3 years of age. She reports that since the start of winter this year, Daniel has been troubled by a dry cough and episodes of laboured breathing. You know from your previous consultations with Maria that she is keen on non-pharmacological therapy where possible. Maria asks for your help with Daniel's cough.

QUESTION 1

What is your initial approach to this consultation?

FURTHER INFORMATION

The history reveals that Daniel has had a cough and a wheeze for 2–3 days and nights each week for the past 2 months. Maria has tried a humidifier in the room but that did not relieve Daniel's symptoms. In desperation, she borrowed a salbutamol puffer from her sister when she was visiting with her daughter, Daniel's cousin, who has asthma. Maria felt the puffer helped Daniel.

Your examination reveals a well-looking 3-year-old boy who is not in any respiratory distress. He does, however, have a bilateral wheeze. His growth parameters are normal for his age.

QUESTION 2

What is your initial diagnostic impression?

QUESTION 3

What differential diagnoses should you consider before making a diagnosis of asthma for the first time?

QUESTION 4

How would you classify the pattern of Daniel's asthma?

QUESTION 5

What treatment might you initiate for Daniel? What instructions would you provide for Maria?

QUESTION 6

Maria is concerned about giving Daniel corticosteroids. How might you address Maria's concerns?

QUESTION 7

Maria asks about alternative and complementary medicines, and non-drug therapies for asthma. What would you tell her?

FURTHER INFORMATION

Maria and Daniel return 4 weeks later for review. Your assessment of Daniel now is that he has shown some improvement but still has symptoms on 1–2 days and nights per week and he uses the reliever twice during the week

QUESTION 8

If spacer technique is good and asthma control remains poor, what might you do next?

CASE 1 ANSWERS

ANSWER 1

It is important to take an appropriate history and conduct a suitable examination. Questions for the history may include:[1]

- How long has Daniel had the cough?
- Is it a daytime or night-time cough?
- Are there any aggravating or relieving factors?
- Is Daniel exposed to any triggers in his environment such as smokers in the house/around the house?
- What is the frequency of the symptoms?
- Are there any associated symptoms such as fever?
- Can Maria clarify the meaning of 'laboured breathing'?
- Has Maria taken Daniel to the hospital at any point? If so, ascertain the outcome and ask to see the discharge summary.
- Has Maria already used any measures to alleviate the symptoms?

The history taking should also include:

- a systems review (eg has there been any diarrhoea, vomiting, snoring, recent upper respiratory tract infection [URTI]?)
- review of growth centiles in the paediatric record book ('Blue book')
- vaccination status
- family history of respiratory or any other medical problems
- travel history.

Examination should focus on the following:[1,2]

- the level of immediate respiratory distress – respiratory rate, subcostal recession, ability to speak in sentences
- colour – pallor, cyanosis
- signs of chronic lung disease such as clubbing
- chest examination – looking for unilateral (asymmetrical) or bilateral (symmetrical) signs such as wheeze, crackles, bronchial breath sounds
- growth parameters and graph on centile charts
- associated conditions (eg atopy).

ANSWER 2

Daniel's history and your examination findings suggest that Daniel may have asthma.

The ideal methodology for diagnosis of asthma is spirometry, but in children under the age of 7 years this may not be achieved reliably. Hence, a history that includes recurrent wheeze, cough or chest tightness associated with a concordant family history of asthma or atopy can be critical in making a diagnosis. A response to bronchodilator therapy can also provide a helpful retrospective confirmation of the diagnosis.[2]

ANSWER 3

Other conditions that can mimic asthma include (but are not limited to):[3]

- inhaled foreign body
- gastro-oesophageal reflux disease (GORD)
- cystic fibrosis
- persistent viral or other infectious disease symptoms such as pertussis or bacterial bronchitis
- tracheomalacia
- habit cough – classically absent when sleeping and treated with suggestion therapy[2]
- sleep apnoea
- hyperventilation and anxiety attacks – these may mimic the shortness of breath of asthma but spirometry during an attack may not show an asthma pattern.
- travel-related conditions (if history of recent travel is given).

Asthma is characterised by variable airflow limitation associated with respiratory symptoms such as wheeze, shortness of breath and chest tightness, which may vary over time.[1]

ANSWER 4

Daniel's asthma could be classified as intermittent, frequent or persistent and, if persistent, as mild, moderate or severe (*Table 1*).[1]

As Daniel has daytime and night-time symptoms on 2–3 days per week, his condition can be classified as moderate persistent asthma.

ANSWER 5

It would be prudent to commence Daniel immediately on a low dose of inhaled corticosteroid such as fluticasone 50 µg twice a day via spacer as a preventer, together with salbutamol 100 µg given as six inhalations via spacer as a reliever (for symptom relief) when required.[1,4,5] Maria should be advised to ensure that Daniel rinses his mouth after inhalation to minimise oropharyngeal candidiasis and systemic absorption of the corticosteroid.[5]

The correct way to use a spacer is to allow 4–6 breaths per puff of the inhaler. Explain to Maria that this can take some time and requires patience, especially with children. For younger children, a mask may be required in order to obtain a good seal,[5] but at the age of 3 years Daniel should be able to manage without one.

An alternative to inhaled corticosteroids is montelukast. The British Thoracic Society guidelines[4] suggest inhaled corticosteroids should usually be the first-line preventer; however, the *Australian Asthma Handbook*[1] allows for the use of montelukast as first-line agents.

You should provide an asthma action plan[6] with instructions on the correct use of a spacer device. Two copies of the asthma action plan should be given to Maria – one for home and one for childcare.

You should also obtain consent for and prepare a GP management plan (Medicare item 721) and give Maria a copy. This document, along with the asthma action plan, should assist Maria in managing Daniel's asthma. You could also direct Maria to the *Australian Asthma Handbook*,[1] which is available online.

Table 1. Definitions of asthma in children aged 0–5 years not taking regular preventer[1]

Category			
Infrequent intermittent asthma	Symptom-free for at least 6 weeks at a time (symptoms up to once every 6 weeks on average but no symptoms between flare-ups)		
Frequent intermittent asthma	Symptoms more than once every 6 weeks on average but no symptoms between flare-ups		
Persistent asthma	**Mild** At least one of: • Daytime symptoms† more than once per week but not every day • Night-time symptoms† more than twice per month but not every week	**Moderate** Any of: • Daytime symptoms† daily • Night-time symptoms† more than once per week • Symptoms sometimes restrict activity or sleep	**Severe** Any of: • Daytime symptoms† continual • Night-time symptoms† frequent • Flare-ups frequent • Symptoms frequently restrict activity or sleep

† Symptoms between flare-ups. A flare-up is defined as a period of worsening asthma symptoms, from mild (eg symptoms that are just outside the normal range of variation for the child, documented when well) to severe (eg events that require urgent action by parents and health professionals to prevent a serious outcome such as hospitalisation or death from asthma).
Note: Use this table when the diagnosis of asthma can be made with reasonable confidence (eg a child with wheezing accompanied by persistent cough or breathing difficulty, no signs or symptoms that suggest a potentially serious alternative diagnosis and the presence of other factors that increase the probability of asthma such as family history of allergies or asthma).
Reproduced with permission from the National Asthma Council Australia from the Australian Asthma Handbook, Version 1.0. Melbourne: National Asthma Council Australia, 2014. Available at www.asthmahandbook.org.au

ANSWER 6

The issue of growth retardation due to inhaled corticosteroids has been the subject of much research over the years. It has been shown that inhaled corticosteroids may lead to a reduction in height of 1 cm in the first year of use.[7] Furthermore, a 2014 Cochrane review[8] involving 10 trials and 3394 children found a small but statistically significant difference in growth velocity in the first year of inhaled steroid use. The magnitude of this effect was deemed to be dose-related: growth velocity was 5.94 cm/year in the low-dose group (50–100 µg beclomethasone equivalent) and 5.74 cm/year in the higher-dose group (200 µg beclomethasone equivalent). However, as no studies in the review went beyond 12 months, the reviewers concluded that, although it would be prudent to minimise inhaled steroid use to the lowest dose for the smallest possible time, further studies are required in this area.[8] A large study that ran for 10 years[9] showed that final adult heights achieved, even after a mean of 9.2 years of inhaled corticosteroid use, were similar in the treatment and the control groups.

It is therefore possible that the growth retardation effect seen with inhaled corticosteroids may be present only at the initiation of therapy and may not persist beyond the first year.[5,9] As with all medications, the decision to treat with inhaled corticosteroids should always be made after weighing up potential benefits versus harms and having a full and frank discussion with the patient.

It would be prudent to explain to Maria in plain English that as with all medications, inhaled corticosteroids do have risks but we would aim to use the lowest possible dose for the shortest time and that given the severity of Daniel's asthma, presently, the benefits would outweigh the risks.

ANSWER 7

You explain that at this stage there is no substantial evidence that any of the following measures can help:[4]

- avoidance of common allergens like dust mite
- modified infant formula
- fish oil
- vitamin E supplementation
- selenium supplementation
- dietary sodium restriction
- magnesium supplementation
- vitamin C supplementation
- probiotics
- acupuncture
- homeopathy
- hypnosis
- chiropractic therapy
- pyridoxine (vitamin B6)[1]

There is mixed evidence for herbal and traditional Chinese medicine and further studies are required to elucidate their role, if any.[4]

You advise Maria that in Daniel's case, as he has moderate persistent asthma, sole reliance on non-pharmacological therapy could prove deleterious to his health and should be avoided until his asthma is brought under control and a 'step-down' can be attempted.[4]

There is good evidence to support avoidance of exposure to cigarette smoke[1,10] and, although not strictly relevant to Daniel, should Maria decide to have another baby, there is some evidence that breastfeeding can have a protective effect as well.[4] Furthermore, while not relevant to Daniel's case, weight loss in overweight or obese patients can help asthma control[4] and cineole (a component of eucalyptus oil) has also been shown to help.[1]

ANSWER 8

If asthma control remains poor, an increase in inhaled corticosteroid dose could be considered and if the response is still inadequate at a dose of 500 µg of fluticasone per day,[5,11] it may be worth reconsidering the diagnosis and referring Daniel to a paediatrician or

paediatric respiratory physician for a second opinion. Referral should also be considered if there are severe asthma exacerbations despite maximal therapy.

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CASE 2

CHARLIE HAS LOOSE MOTIONS

Charlie is brought to see you by his mother. Charlie is 3 years and 9 months of age. His mother reports that Charlie has had loose motions over the last 3–4 months. She is worried about the possible causes.

QUESTION 1

What key parts of the history do you need to ask about?

FURTHER INFORMATION

You learn that Charlie was born at term weighing 3.25 kg after an unremarkable pregnancy and was breastfed for the first 7 months of life. Charlie was well as an infant and his growth and developmental progress were normal. However, he was hospitalised with diarrhoea and vomiting just after his third birthday. Although not substantiated at the time, these symptoms were thought to be secondary to a viral infection. The symptoms settled over 7–8 days. Charlie lost some weight at the time of the acute diarrhoea (about 700 g), but regained this weight within 1–2 weeks. Subsequently, however, his mother thinks that he has not gained much more weight. He now weighs 14.5 kg.

He has been more lethargic and grumpy than usual in the past few months, and reports some vague abdominal pain most days. He has up to four loose unformed bowel motions each day, but no urgency and no blood.

Charlie has no other past medical or surgical history. There is no history of travel, or known contact history. Charlie has no siblings. His father has well-controlled type 1 diabetes mellitus (T1DM) but is otherwise well. The only other family history of possible relevance is that his mother's sister has what she calls irritable bowel syndrome (IBS).

QUESTION 2

What are the key aspects of examination that you will focus on?

FURTHER INFORMATION

You arrange to measure Charlie's weight and height, and plot these on an appropriate growth chart. You compare these to his previous measurements and note that his height has continued along the 50th percentile for age. By contrast, Charlie's weight, which was at the 50th percentile before his third birthday, is now between the 10th and 25th percentiles for age.

You complete an abdominal examination and find no specific abnormalities. There is no distension, no organomegaly and no presence of mass. There is no tenderness and bowel sounds are normal. You do not note any pallor or oedema.

QUESTION 3

What are the likely differential diagnoses for Charlie's symptoms? What tests would you perform?

QUESTION 4

What would you recommend for Charlie while you await the results of these investigations?

FURTHER INFORMATION

The results, shown in *Table 1,* indicate low iron stores and a microcytic anaemia. C-reactive protein (CRP) and albumin are both normal, and stool microscopy and culture show no white cells and no pathogens. Tissue transglutaminase antibody (tTG) and deamidated gliadin peptide (DGP) levels are elevated.

Table 1. Test results

Test	Charlie's result	Normal ranges
Stool microscopy and culture	Normal	
Full blood evaluation (FBE):		
• Haemoglobin	103 g/L	120–140 g/L
• Mean corpuscular volume (MCV)	77 fL	78–93 g/L
Iron studies:		
• Ferritin	9 mg/L	20–200 mg/L
Albumin	Normal	
CRP	Normal	
Coeliac serology:		
• IgA tTG	78 IU	<5* IU
• IgG DGP	62 IU	0–20* IU
• Total IgA	0.5 IU/mL	0.3–0.7 IU/mL

*Normal ranges vary with different test kits

You refer Charlie to the local paediatric gastroenterology service for small bowel biopsy (endoscopic biopsy from proximal small bowel). This is completed 3 weeks later.

The endoscopy report indicates scalloping of the duodenal folds, with several small erosions in the duodenal cap. The histology results are available 4 days later and indicate the presence of crypt hyperplasia, patchy villous atrophy and intraepithelial lymphocytosis. No other changes are evident.

QUESTION 5

How would you interpret these results?

QUESTION 6

What would you recommend for Charlie and his family?

QUESTION 7

How would you follow his progress?

FURTHER INFORMATION

At a subsequent visit, Charlie's mother, who screened negative for coeliac disease, reports that she is pregnant with their second child. She asks about her diet during pregnancy, when to introduce gluten into her new baby's diet and when and how to screen for coeliac disease.

QUESTION 8

What will you tell Charlie's mother?

CASE 2 ANSWERS

ANSWER 1

You will need to ascertain the presence of any associated symptoms. Several of the conditions included in the differential diagnosis for Charlie are likely to be familial in nature and so documenting family history will be important.

ANSWER 2

It is particularly important to assess nutritional consequences of chronic diarrhoea in a toddler.[1] These include weight gains and linear growth.

Assessment of growth is an important aspect in the examination of children with chronic gastrointestinal symptoms. Although the current growth parameters are important, these need to be seen in the context of historical growth data (what has happened over time?). The relationship between height and weight is also a key aspect of interpretation.

Explore nutritional aspects, including looking for micronutrient deficiencies; the most common is iron deficiency. Others include vitamin D, vitamin B12 and folate deficiency.[2]

Warning signs of significant mucosal disease might include the presence of blood (haematochaezia), significant weight loss or slowing of weight gains, repeated hospital admissions requiring fluid support, and nutritional findings (eg oedema secondary to enteric protein loss).

ANSWER 3

Possible differential for these symptoms include:[1]

- gastrointestinal infection, such as from parasites (giardia or strongyloides)
- coeliac disease
- constipation with overflow
- carbohydrate intolerance (eg lactose intolerance or excessive ingestion of fructose)
- inflammatory bowel disease (IBD)
- small intestinal bacterial overgrowth.

Appropriate initial investigations would include:[1,2]

- stool microscopy and culture, with specific request for parasites
- FBE
- iron studies, including ferritin levels
- albumin
- CRP
- coeliac serology (IgA tTG, IgG DGP) antibody, total IgA.*

*The total IgA level is often included automatically, depending on the test kit used by the laboratory and local laboratory practice. Standard IgA tTG will not be reliable in individuals with IgA deficiency, in which case an IgG-based serological test, such as IgG tTG or IgG DGP, is required. Also, IgA tTG tests may not be as reliable in young children.

ANSWER 4

Given that the differential diagnosis includes lactose intolerance, it would be reasonable to arrange a food and symptom diary. This could establish connections between ingestion of dairy foods (containing lactose) and symptoms.

If there appears to be a relationship between dairy products and diarrhoea, it may be reasonable to suggest a short period of lactose exclusion (over 7–10 days). All obvious dairy products should be excluded and replaced with lactose-free alternatives (lactose-free cow's milk or soy products). Clearly, dairy products provide an important source of calcium and should not be excluded indefinitely without clear indication and without consideration of calcium intake.

Similarly, if Charlie seems to be having a large volume of undiluted fruit juice every day, it would be appropriate to recommend a reduction in this intake.

Although coeliac disease is on the differential list, an empirical trial of a gluten-free diet should not be undertaken at this time.[2] Given that the management of coeliac disease requires a life-long dietary change, it is essential to ensure that the diagnosis is established definitively at this stage. An empirical trial of gluten exclusion or reduction may lead to partial mucosal improvements, which then interferes with the interpretation of the duodenal histology, thereby preventing or delaying a definitive diagnosis.

ANSWER 5

The endoscopic and histological features reported are consistent with and diagnostic of coeliac disease. It is important to recall that confirmation of coeliac disease requires duodenal biopsy.[3,4] Given Charlie's presentation with gastrointestinal symptoms, his positive coeliac serology and his biopsy results you have no doubt that he has coeliac disease.

Even just a few decades ago, most children diagnosed with coeliac disease presented with so-called 'classic coeliac disease'. This pattern involved severe malabsorptive symptoms and significant

failure to thrive. Although some toddlers still present in this manner, most children now present with just one or two gastrointestinal symptoms or even with non-intestinal symptoms. Preschool children more often have diarrhoea and interruption to growth, whereas older, school-age children more commonly present with abdominal pain.[5,6]

ANSWER 6

You should arrange to discuss the histology results with Charlie's parents. You should discuss the aetiopathogenesis of coeliac disease, the key elements being diet, at-risk genes and the immune response.[7,8] Coeliac disease is initiated by the toxic component of gluten proteins (from cereals), which we are unable to digest. This protein moiety is able to enter the small intestinal mucosa where it is processed and then detected by antigen-presenting cells. If these cells contain the correct human leukocyte antigen (HLA) molecules, reflecting genetic risk, then these activated cells are able to stimulate a T lymphocyte response. This immune response leads to the mucosal changes that were seen on Charlie's biopsies.

Charlie should now commence a gluten-free diet, which will be a life-long dietary change. You send off a referral to the local paediatric dietetic service requesting dietetic education for the family. You also provide the contact details for Coeliac Australia and the local support group (for support, resources and information updates) and provide written confirmation of the diagnosis, which is required for Coeliac Australia membership.[2,4]

Given the increased risk of coeliac disease in first-degree family members (approximately 10% in first-degree family members[2,7]), you should also arrange for Charlie's parents to have coeliac serology bloods completed.[2,4] His father has additional risk as there is an association between coeliac disease and T1DM (5–10%).[3] You recall the family history of Charlie's aunt and you mention to his mother that the aunt may also have coeliac disease and this may be contributing to her IBS symptoms.

ANSWER 7

In terms of follow-up, check on Charlie's initial adjustment to a gluten-free diet after a few weeks. At this time it may be reasonable to add some iron supplements to help build up Charlie's iron stores (if he has features of anaemia on history and/or examination).[2,3]

Charlie should be reviewed 6 months after diagnosis to check on his adherence to the gluten-free diet, and to re-assess his growth (expecting that he has regained weight at this time), check on resolution of symptoms and repeat examination. You should arrange for repeat serology tests, and for repeat FBE and iron status.[2,3]

Subsequently, Charlie should be reviewed annually.[1–3] At these times, you would check his symptoms, document his weight, review his adherence to the gluten-free diet and arrange monitoring blood tests, including coeliac serology. At these times, you should remind Charlie and his parents of the importance of adherence to the gluten-free diet. Full adherence is important in terms of preventing recurrence of symptoms, ensuring maintenance of normal duodenal mucosa and preventing long-term complications of untreated coeliac disease. These include gastrointestinal complications (eg diarrhoea, pain, lymphoma) and non-gastrointestinal complications (eg osteoporosis, iron deficiency, poor growth, dental enamel and infertility).

ANSWER 8

Background

Coeliac disease is clearly associated with HLA types DQ2 and DQ8.[3] The absence of these genes effectively excludes the risk of coeliac disease occurring in the future. However, although HLA positivity is essential, it is not sufficient for the development of coeliac disease, as the majority of people with these HLA types do not develop coeliac disease, and other genes and other factors must be involved. Up to 50% of the Australian and New Zealand populations have one or both HLA types,[7] whereas the prevalence of coeliac disease is in the order of 1–1.5%.[2,8]

Evidence suggests that the preferred time to introduce gluten is at 4–6 months of age to maximise the potential for immunological tolerance in genetically predisposed individuals.[9] Breastfeeding at this time was thought to be additionally protective but this has been questioned by recent research.[8,10]

It may take several years from the introduction of gluten for positive antibodies to develop and results may be falsely negative even in children under 2–4 years of age with symptoms.

Advice for Charlie's mother

As Charlie's mother has already been screened and found not to have coeliac disease, there is no need for her to be on a gluten-free diet and there is no evidence to suggest that this would in any way alter the risk of her child developing coeliac disease.

Advice about the age at which to introduce gluten-containing solids should be the same as for other infants and, in particular, Charlie's mother should be advised not to deliberately delay their introduction.

Should the new baby go on to develop symptoms suggestive of coeliac disease, it is important to advise her not to institute a gluten-free diet before a firm diagnosis has been made by blood tests and biopsy. In the absence of symptoms, expert opinion recommends routine screening at about ages 4, 7 and 12 years.

It is also possible to test the new baby's HLA status. This can be done on a saliva sample or buccal smear in young children, avoiding the need for blood tests. If negative, then no further screening or monitoring steps are required. If positive for DQ2 and/or DQ8 then screening must be carried out as above.

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CASE 3

CHRIS HAS DISTRESSING PAIN IN THE TUMMY

Chris, aged 6 years, attends with the whole family. Chris looks well today but his mother Jill is upset. She explains that last night Chris had a bout of tummy pain and says, 'He was screaming and writhing around on the floor. I was really frightened so I took him to the emergency department but there was a 4-hour wait'. After 1 hour in the waiting room, Chris was back to his normal self so Jill decided to take him home before the doctor saw him. She then made an urgent appointment to see you this morning. She tells you, 'It has been going on for months … far too long'.

QUESTION 1

Having listened to this story and observed Chris and his family as they settled in to your consulting room, what goals for this consultation are you developing?

FURTHER INFORMATION

Your empathic approach works well and Jill's concern dissipates as the consultation progresses. She appreciates your thorough questioning and the rapport you are building with Chris.

QUESTION 2

At this early stage, what is your provisional diagnosis? What differential diagnoses would you consider?

FURTHER INFORMATION

Chris says he has to sit on the toilet for quite a long time and that his poo is often hard. Jill adds, 'Another thing, doctor, he's had diarrhoea lately and has come home from school with dirty underpants quite a few times recently'.

QUESTION 3

What are the implications of this new information for your provisional diagnosis?

QUESTION 4

How will you confirm your provisional diagnosis?

FURTHER INFORMATION

Chris gets more comfortable as the examination proceeds. You ask him, 'Is it ever painful when you are actually pushing out your poo?' He says, 'No'. You ask, 'Do you ever use the toilet at school Chris?' He says, 'Oh yes'.

QUESTION 5

Why is this information helpful?

QUESTION 6

What, if any, investigations are indicated?

QUESTION 7

What else should your consider in this consultation?

QUESTION 8

What plan of management will you propose for Chris and his family to consider?

CASE 3 ANSWERS

ANSWER 1

Appropriate goals for this consultation would be to:

- win Chris's mother's trust and settle her distress
- take a thorough history and perform an appropriate physical examination
- develop a provisional and differential diagnosis for Chris
- involve Chris and his family in agreeing to a management plan.

Be clear about the importance of achieving a therapeutic relationship with Chris and especially with his mother. Experience suggests that failure to involve Chris and his family in agreeing to a management plan often results from failure to win the parents' trust. In this case, Chris's mother needs the doctor to listen to her, hear and validate her concerns, and contain her distress.

Examples of empathic statements that experience suggests are likely to be helpful include:

- 'That was really upsetting.'
- 'A horrible night ... '
- 'It's really hard to watch when a child is in such pain – you feel so helpless ... '
- 'No one likes to see their children in distress.'

ANSWER 2

On the basis of your observations of this apparently well boy with a long history of episodic, distressing abdominal pain, idiopathic constipation (meaning there is no physiological or anatomical cause), is the most likely diagnosis.[1] However, it is important to obtain a thorough history and perform a physical examination to confirm the diagnosis.

Other causes of episodic abdominal pain that might include or be confused with constipation include Hirschsprung's disease,[2] which is most likely to present in the first weeks of life with delayed passage of meconium, abdominal distension and repeated vomiting[3]. A child with intussusception[4] or volvulus[5] can look remarkably well in between episodes of pain. Intussusception is the most common cause of intestinal obstruction in children up to 3 years of age.[4] Volvulus commonly presents in the early months of life but either condition can occur in older age groups.[4,5]

Other, non-idiopathic causes of constipation are exceedingly rare but need to be kept in mind. They include:

- coeliac disease
- hypothyroidism
- neurological dysfunction with lower limb signs
- an anteriorly placed anus.[2,6]

ANSWER 3

The loose stool of encopresis is commonly mistaken as diarrhoea.[1,2,6] This is one of many reasons that Jill might find it hard to share the provisional opinion that constipation is the underlying cause of her son's pain. You explain that this 'pseudo-diarrhoea' is in fact loose stool seeping around a dam of impacted faeces. The 'diarrhoea' will be fixed by treatment of the underlying constipation, although that may seem counterintuitive.

ANSWER 4

You should obtain a more complete history and perform a physical examination.[2]

It can be useful, early in the consultation, to establish if Jill has a view on what is causing the problem. Features in the history that suggest idiopathic constipation include recurrent abdominal pain, lengthy periods sitting on the toilet trying to pass stool, stools that are hard in texture and reduced frequency of passing stools. There may be a history of constipation in the past. It is important to establish if the actual passage of stool through the anus is painful. In young children there may be a history of behaviours that display efforts to delay the passage of stool. There may be a history of 'pseudo-diarrhoea' or encopresis as discussed above. Constipated children may have reduced appetite and may have become irritable or less joyful. Does the patient usually have a healthy diet? An appreciation of the social context and family functioning are important here as in most consultations with children.

Check for a softly distended abdomen with palpable faeces in the left lower quadrant and increased bowel sounds, including tympanic sounds. If impaction has been present for any length of time, each of these signs will be more pronounced. Inspecting the anus might reveal an anal fissure or perianal streptococcal infection. It is important to check that height and weight velocities are normal.[2] The National Institute for Health and Care Excellence (NICE) guideline[2] recommends inspection of the skin and anatomical structures of the lumbosacral/gluteal regions, and ensuring that there is a normal gait and normal tone and strength in lower limbs. Experience suggests that an appropriate history and examination all but confirm a diagnosis of idiopathic constipation. Other, much rarer, possibilities in the differential diagnosis need to be kept in mind as follow-up continues.

Digital rectal examination should only be undertaken, if at all, by an experienced clinician who is able to interpret the findings.[2]

ANSWER 5

Lack of pain during anal dilation makes an anal fissure and other perianal conditions, such as streptococcal infection, unlikely. It also makes fear of the feeling of anal dilation unlikely to be a causal element in this presentation. You have eliminated the quite common fear of using toilets at school, which, if present, might arise from a spectrum of issues ranging from family culture through past episodes of bullying in the toilets, and on to anxiety disorders. Pseudo-diarrhoea with encopresis favours your provisional diagnosis and suggests impacted faeces.[2]

ANSWER 6

No clinical investigations are indicated.[2] Specifically, the NICE guideline[2] advises against the use of use a plain abdominal radiograph or abdominal ultrasound to diagnose idiopathic constipation. Normal growth velocities make hypothyroidism or coeliac disease unlikely causes for Chris's constipation.

ANSWER 7

As the consultation proceeds, you should check that Jill's understanding of the problem is developing as you hope it is. Many parents find it hard to accept that the agonising episodes of pain they have observed in their children, and failed to relieve, could possibly be caused by something as 'simple' as constipation. 'He has a really good diet', they often say. If Jill is resisting your explanations then stop and declare it, 'I suspect you aren't convinced?' The following silence, and then her response, may provide clues that will allow you to empathically explore the reasons for her lack of conviction. These must be dealt with if the consultation is to succeed.

A 'script' that might help when parents' doubts persists is something like:

> 'These pains are horrible to have and horrible to watch. They can be excruciating (or, they are really, really horrible ... they really, really hurt). On the other hand, they are not dangerous ... they are not risky for Chris's health ... but they are horrible and we can all work together to get rid of them'.

ANSWER 8

Non-pharmacological components include:[1,2]

- maximising parental confidence and competence
- minimising any environmental stressors impacting on Chris (including toileting strategies)
- encouraging healthy eating, drinking and exercise.

Chris has impacted faeces. Features suggesting impaction include chronicity, easily palpable faecal masses and encopresis.[2] In this case, it is necessary to disimpact first. An oral approach to disimpaction is preferred.[1,2] This is usually achieved at home with increasing doses of Movicol.[1] In Australia, one of the proprietary formulations of polyethylene glycol 3350+ electrolytes is licensed for disimpaction and maintenance in children from 2 years of age.

Readers should consult their usual formularies for disimpaction and maintenance regimes. The table of laxatives and doses in the Royal Children's Hospital Guideline is easily accessed, clear and concise.[7] The guidelines[1,2,6,7] offer advice on adding other laxatives if needed. There is only one macrogol approved for use in children as young as 2 years of age, which comes in two paediatric formulations, one with flavouring and one without. Unlike the adult formulations, the paediatric formulations are not listed on the Pharmaceutical Benefits Scheme (PBS) for chronic constipation. The most common causes of failed treatments are inadequate doses and failure to persist for an appropriate length of time.[1]

The parents and child need to know that this initial phase of treatment is likely to be messy and painful. It should happen at home and will need time off from pre-school or school. The disimpaction regime should cease when Chris passes a large quantity of stool or watery diarrhoea. He should then be transferred to a maintenance regime (described below).

It can be helpful to see Chris at least weekly to check progress, maintain morale and to encourage persistence with the maintenance regime. Maintenance usually needs months rather than weeks to be effective and requires daily use of laxatives recommended for children.[1,2,5] It is important to advise parents that there is no evidence that long-term laxative use is harmful, addictive or damaging.[5] Families may need a lot of encouragement to persist.

RESOURCES FOR PATIENTS

- Raising Children Network has a variety of essays on constipation and related subjects aimed at parents, http://raisingchildren.net.au/articles/constipation.html/context/555

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CASE 4

XAVIER IS VOMITING AFTER FEEDS

Xavier is 5 weeks of age. The maternal child health nurse asks you to see him because his mother, Susie, complains that he is very unsettled and vomits frequently. He is being breastfed and when his mother fed him at the appointment his sucking was very noisy. He did not cry during the feed or arch or pull off and finished feeding from both breasts. However, 5–10 mins after his feed he brought up a large amount of undigested milk.

QUESTION 1

What features would be important to ask about in the history?

FURTHER INFORMATION

Xavier is Susie's second child. She had an uneventful pregnancy and delivery, and Xavier was born vaginally at 39 weeks. He weighed 3.68 kg at birth and had Apgar scores of 9 at 1 minute and 5 minutes. It took 5 days before Susie's breast milk came in and Xavier lost 10% of his birth weight. He is now gaining weight and has put on 300 g in the last week. He is on the 50th percentile for weight and height on his growth chart. Xavier is fed on demand every 2–3 hours and twice overnight. Susie reports that Xavier cries when he is lying on his back and does not settle unless held upright and rocked. He likes to be held and patted before sleeping but sleeps well between feeds. He gulps a lot when feeding but does not cry or arch and completes a full feed. Initially, Susie had considerable nipple pain when feeding but she saw the hospital's lactation consultant who helped with Xavier's latch, which has now improved and Susie's nipple pain is getting better. Xavier passes 4–5 stools a day but they are not green or frothy. He is not crying when you see him and looks comfortable in Susie's arms.

Susie had postnatal depression with her first baby, as well as breastfeeding problems that required intensive support with a lactation consultant and GP for the first 3 months.

Note, the average birth weight of most babies is approximately 3–3.5 kg and it is not uncommon for babies to lose weight in the first few days through loss of prenatal fluid.[1] Weight loss of <10% is considered acceptable but >10% suggests that there is a problem with feeding or absorption. Babies should regain their birth weight by their second week of life and continue to gain 100 g or more each week in the first 3 months.[1] Most breastfed babies feed 8–12 times in a 24-hour period, including 2–3 feeds overnight.[1]

QUESTION 2

What features should be examined?

FURTHER INFORMATION

Xavier is well hydrated and has a normal fontanelle. His facial features and palate are normal. His pupils and red reflex are normal. He has two heart sounds, no murmurs or added sounds, and his chest is clear. His abdomen is soft and there are no masses, and his umbilicus has healed well. His hips and genitalia are normal, and his neurological examination is normal. His skin is normal and there are no signs of eczema.

During your discussion and after your examination Xavier falls asleep in his car carrier.

Susie's mood is good and she is enjoying her baby more this time but is concerned about Xavier's crying and vomiting. She says, 'The maternal child health nurse was concerned that this might be reflux. He does vomit a lot. He can cry quite a bit at times and I can't hold him all the time as I have his older sister to take care off. Is there anything I can do to stop his vomiting?'

QUESTION 3

What are possible diagnoses for Xavier's vomiting?

QUESTION 4

What is gastro-oesophageal reflux (GOR)?

QUESTION 5

What features would make you suspect gastro-oesophageal reflux disorder (GORD) instead of simple regurgitation?

QUESTION 6

What management options would you suggest for Xavier's vomiting and crying?

CASE 4 ANSWERS

ANSWER 1

A general neonatal history should be taken from the mother, including asking about:

- the pregnancy, delivery and any special care needed immediately after the birth
- Xavier's weight and signs of normal developments
- further details about fever and regurgitation/vomiting including:
  - the colour of the vomit (to determine if there is blood or bile in the vomit)
  - the timing of episodes
  - amount of regurgitation/vomiting
  - the force involved
- other associated symptoms.

A feeding history should be elicited, asking:

- How often does the baby feed?
- If formula-fed, what amounts does the baby take?

It is important to know if there have been any difficulties with breastfeeding and questions should be asked about the baby's crying, settling and sleep.

Additional questions include:

- How is the mother is coping?
- Are there are other siblings who need care?
- What sort of support does the family have?
- Is there is any history of mental health problems?

ANSWER 2

A full examination should be carried out including:

- body temperature measurement
- assessment of hydration
- fontanelle and palate examination
- abdominal examination
- genitalia and anus examination
- assessment of skin
- examination for normal spine
- neurological examination.

If there are still concerns about breastfeeding, it can be useful to observe a feed, looking for a good latch and feeding technique, as well as any signs of nipple damage.

Xavier's growth charts and his hand-held care record should be reviewed.

Xavier's weight should be measured, using specific baby scales, and this should be done without clothes or a nappy.

ANSWER 3

Xavier has signs of regurgitation with some crying and irritability, but with normal growth patterns. The most likely diagnosis is a normally growing and developing baby with physiological gastro-oesophageal reflux (GOR). However, it is important to exclude other causes of vomiting in infants[2] and exclude red flags.[3]

Other gastrointestinal disorders include:

- GORD: symptoms may include vomiting, feeding refusal and failure to thrive
- pyloric stenosis: projectile vomiting
- malrotation: bilious vomiting with abdominal distension
- cow's milk protein allergy (CMPA): vomiting, diarrhoea, eczema and urticaria linked to exposure to cow's milk and can occur with formula or breast milk
- hepatitis: jaundice and right upper quadrant pain
- viral gastroenteritis: vomiting, diarrhoea and fever.

Urinary tract infections can also cause vomiting.

Meningitis and hydrocephalus should be excluded.

Metabolic disorders, such as renal tubular acidosis, urea cycle defects and hypocalcaemia, are rarer causes of chronic vomiting in infants and children.[4]

ANSWER 4

GOR is the passage of gastric contents into the oesophagus (with or without regurgitation and vomiting) lasting <3 minutes in the postprandial period, with few or no symptoms. It is the result of a laxity in the lower oesophageal sphincter, which in infants is due to developmental immaturity. Infantile GOR peaks at 4 months of age[5] and is usually improved[1] or resolved by age 12 months.[6] Around 70–85% of infants have regurgitation within the first 2 months of life.[7] GOR is benign and does not impact on the baby's health.[2]

ANSWER 5

GORD is suspected when GOR causes troublesome symptoms and or complications[8] such as:

- failure to thrive
- haemtemesis
- refusal to eat
- aspiration pneumonia
- sleeping problems
- chronic respiratory disorders
- oesophagitis
- stricture
- anaemia
- apnoea.

Complications are serious but uncommon.[8] Referral to a specialist should be sought for further investigation if the above symptoms are present.[1,8]

GOR may be the presenting features of food allergy, most commonly to dairy products and sometimes to soy.[2] CMPA is an immunologically mediated adverse reaction to cow's milk protein. It can present as a range of symptoms occurring from minutes to hours to several days after ingestion of cow's milk formula.[8] Children with CMPA are often irritable, may have diarrhoea and may demonstrate feeding refusal.[2] Up to 40% of infants with symptoms of GORD referred to specialist services are thought to have CMPA.[8] The key element to note on history taking is that symptoms are most likely to develop within 4 weeks of exposure to cow's milk formula.[9] Note that CPMA is not limited to formula-fed infants and can occur in exclusively breastfed infants, as intact cow's milk proteins can be secreted in breast milk.[10]

ANSWER 6

As GOR is transient, the aims of management are to be supportive, reassure the parents that GOR is not harmful and provide parents with good patient education (refer to *Resources*). Providing explanations about the laxity and immaturity of the gastro-oesophogeal junction can be helpful. Highlighting ongoing growth and using the growth charts as a visual aid to reassure patients can also be useful.

Putting the infant in the prone position after feeding can also decrease the incidence of reflux but this should always be done when the infant is awake and with adult supervision.[11] Thickening feeds, for example by using rice cereal, corn starch or commercial food thickeners is often recommended for formula-fed infants,[2,11] but a Cochrane study found no evidence of efficacy, as there was a lack of good-quality randomised controlled trials.[12]

A small study suggested that avoiding exposure to tobacco smoke, avoiding overfeeding, avoiding aerophagia (swallowing of air) by ensuring good attachment when breastfeeding and/or using a bottle effectively led to improvement in GOR symptoms.[13]

Provision of information about settling and the normal period when babies are more likely to cry and be unsettled can also be helpful for parents (refer to *Resources* section)

Infants diagnosed with GOR and GORD are often treated with proton-pump inhibitors (PPIs) or other anti-secretory medicines. However, a recent systematic review of clinical trials showed that GORD is rarely a cause of excessive crying or irritability in infants and PPIs are no better than placebo at relieving symptoms.[14] In infants, gastric acid is buffered for 2 hours after feeding with breast milk or formula and it is thought that the buffered refluxate does not irritate the oesophageal mucosa.,[15] Although short-term use of PPIs seems to be well tolerated, evidence to support long-term safety is lacking.[11,14] If there is no improvement and reflux symptoms have a longer duration, or are causing significant feeding and settling difficulties, or are associated with complications, a trial of PPI therapy may be indicated.[1] Granules or rapidly dispersible tablets may be easier to use in infants. A suitable regimen would be:[2]

- esomeprazole granules, 0.4–0.8 mg/kg (maximum 20 mg), dispersed in water and given orally once daily
- rapidly dispersible lansoprazole tablet, 1.5 mg/kg (maximum 30 mg), placed on the tongue to dissolve, once daily

- pantoprazole granules, 1 mg/kg (maximum 40 mg), dissolved in water or mixed with soft foods, if applicable, and given orally once daily.

There is insufficient evidence to support the use of prokinetic drugs (eg metoclopramide, domperidone or cisapride) and their side effect profile also makes them unsuitable.[2]

In children with CPMA-induced GORD, it is reasonable to eliminate dairy products and to trial a change in formula. For infants older than 6 months, this may include use of infant soy for 2 weeks.[8] If there is no improvement, specialist referral may be warranted to assess if an extensively hydrolysed formula is required.[8] Breastfed infants should not be changed to formula.[11] An elimination diet for the mother can be trialled for 2–4 weeks and if symptoms improve, cow's milk protein can be reintroduced. If symptoms recur, the mother would need to resume the elimination diet.[16] This should be done with support from a paediatric dietician.[16] Non-invasive breath tests for lactose intolerance can be used but accessibility can be difficult and stool-reducing substances are unreliable and non-specific for lactose intolerance and are no longer recommended.[8]

Surgical treatment is reserved for infants in whom medical therapy has failed to control symptoms or recurrent aspiration infections. This requires a specialist review.[8]

RESOURCES FOR PATIENTS

- The Royal Children's Hospital website provides fact sheets on GOR for parents, www.rch.org.au/kidsinfo/fact_sheets/Reflux_GOR/
- The Period of PURPLE Crying website has information about crying in early months and advice on settling, http://purplecrying.info
- Australian Society of Clinical Immunology and Allergy provides an advice sheet on cow milk avoidance diet, www.allergy.org.au/images/pcc/ASCIA_PCC_Dietary_avoidance_cows_milk_soy_2014.pdf
- National Institute for Health and Care Excellence guidelines Gastro-oesophageal reflux disease: recognition, diagnosis and management in children and young people [NG1] provides patient information, www.nice.org.uk/guidance/ng1/ifp/chapter/about-this-information

RESOURCES FOR DOCTORS

- Therapeutic Guidelines Gastrointestinal Therapeutic guidelines – Gastro-oesphageal Reflux in Children
- The Royal Children's Hospital, Melbourne. Gastro-oesophageal reflux guidelines for clinicians, www.rch.org.au/clinicalguide/guideline_index/Gastrooesophageal_Reflux_in_infants
- National Institute for Health and Care Excellence guidelines – Gastro-oesophageal reflux disease: recognition, diagnosis and management in children and young people [NG1], www.nice.org.uk/guidance/ng1

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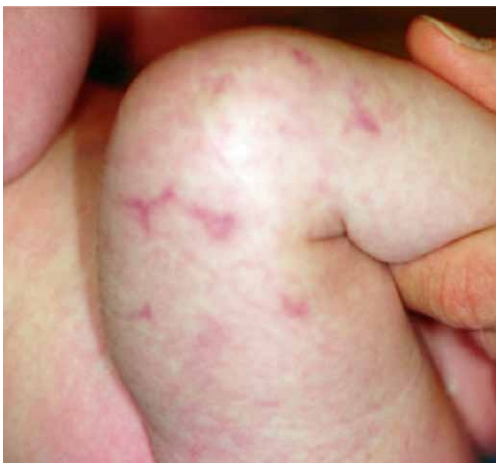
CASE 5

BILLY HAS MARKS ON HIS LEG

Kylie presents with her son, Billy, who is 6 weeks old and has no known health issues. She has come to discuss Billy's immunisations.

The practice nurse undresses Billy and notices what appears to be an unusual rash on his leg (*Figure 1*). Kylie says she noticed the marks a couple of days ago, but does not know what caused them.

Figure 1. Markings on Billy's leg



Reproduced with permission from Springer from Giardino AP, Lyn MA, Giardino ER, editors. A practical guide to the evaluation of child physical abuse and neglect. New York: Springer; 2010.

QUESTION 1

What is your initial assessment of the 'rash' on Billy's leg?

QUESTION 2

What would you do next?

FURTHER INFORMATION

Kylie says Billy has seemed out of sorts the last few days. She had left Billy with her partner or with the neighbours during the past 3 days and she is sure they would have let her know if there were any problems. She says she will go home and talk to the neighbours again. Kylie says she saw the paediatrician last week and there were no concerns. She offers go back to the paediatrician again if you recommend it. Kylie seems very reasonable in her approach and does not seem depressed.

QUESTION 3

Are Kylie's suggestions a reasonable course of action?

QUESTION 4

Can this case be managed and resolved in a general practice setting in a few days?

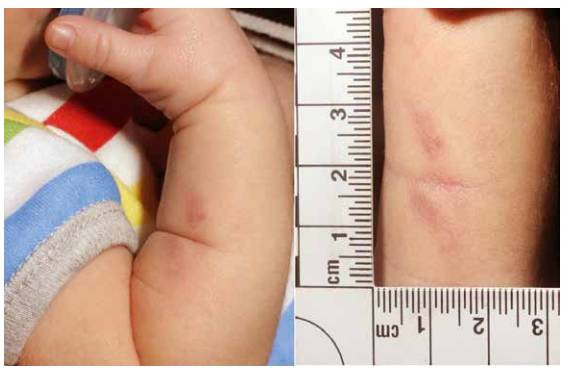
QUESTION 5

Is bruising on the leg of this baby pathognomonic of child abuse or a bleeding disorder?

QUESTION 6

If Billy presented, instead, with two parallel linear bruises 1.5 cm in length on his forearm as shown in *Figure 2*, how would you interpret this?

Figure 2. Red bruising to the forearm of a breastfed baby



Reproduced with permission from BMJ Publishing Group Ltd from Venkata RN, Woolley C. Infantile sucking bruises. Arch Dis Child; 2014

QUESTION 7

What are the most common sites of accidental childhood bruising?

QUESTION 8

What are the uncommon sites for accidental bruising that should raise suspicion?

CASE 5 ANSWERS

ANSWER 1

The red marks shown in *Figure 1* have the appearance of bruises (ie blood in the skin or visible through the skin). It is easier to detect bruising on a person with light coloured skin. The marks have the appearance of grab-marks but for clinical decision-making it is sufficient to note that the child has bruises on his leg.

ANSWER 2

Appropriate next steps include:

1. Explore the history. Ask Kylie if she can remember anything about the marks. When did she first notice them? How has Billy been in terms of his health, feeding, sleeping and mood in the last few days? Record exactly, preferably using her words, any account that Kylie gives of what happened. Also record details of who has been caring for Billy recently.
2. Review any psychosocial information the practice holds about the family and about this child's health.
3. Keep any information you give Kylie general. For example, you could state, 'These marks could be bruises and it is important to check that Billy is well and has no bleeding tendency or problems, and to work out what might have happened'.

These recommendations are based on the dual principles of communication in general practice of giving warning before breaking bad news and, in potentially serious matters, of not contaminating a police investigation.

ANSWER 3

This is not a suitable plan of action.

Any bruising in a baby who is not mobile is highly suspicious and must be comprehensively investigated by a paediatric child protection team. As a mandatory reporter, you must immediately contact the Child Protection Helpline for your state or territory (refer to *Resources*).[1]

The principles of response are:[2]

1. identify (the risk)
2. assess/consult
3. notify
4. manage.

ANSWER 4

This case cannot be managed and resolved in a general practice setting in a few days.

Billy may have other injuries and he requires a paediatric assessment, which may include a full assessment of the circumstances of his injury, a skeletal survey, a scan for subdurals and a retinal assessment.

Billy will probably need to be admitted to hospital for investigations, which also provides time for a police investigation and community services assessment. Admission to hospital is much less confrontational and/or distressing than removal of Billy from his parents while investigations are made. Bruising may also be a sentinel injury noted prior to a diagnosis of child abuse.[3]

Ideally, a parent would be supportive of a hospital admission to investigate the problem. It is important that the child is in a safe place and parental contact is unlikely to be prevented at this stage. Kylie is still breastfeeding and should be supported to continue this while Billy is in hospital.

In children, the age group most likely to require hospitalisation due to assault, are children under 1 year.[3] Infants are at the greatest risk of inflicted traumatic brain injury[4] and death by assault.[5]

If abuse has occurred, Kylie may be involved, with or without mental illness or coercion due to partner violence. Alternatively, Kylie may have no knowledge of what has happened to Billy or she may not have been present at the time of the injury and may believe lies told by the perpetrator.

ANSWER 5

No injury is pathognomonic (characteristic of a particular condition) of child abuse but for certain patterns of injury the probable or very possible explanation is assault. Bruising in non-ambulant infants remains very suggestive of physical abuse and explanations provided by parents should be considered carefully.[6] Bruising may also be a sentinel injury noted prior to the diagnosis of child abuse. This situation requires an assessment led by a child abuse team. Babies and toddlers who fall off the bed, change table or out of the cot, would rarely sustain a moderate injury and will not usually bruise.[7]

According to the Cardiff Child Protection Systematic Reviews, injury in a child 'must never be interpreted in isolation and must always be assessed in the context of medical and social history, developmental stage, explanation given, full clinical examination and relevant investigations'. They also recommend appropriate investigation of any unexplained injury identified in a child that causes healthcare professionals concern.[8] The Royal Children's Hospital, Melbourne recommends that examination should include height and weight measurment; examination of the head, mouth, eyes, ears, chest, abdomen, back and limbs; and the use of body diagrams and a 'physical abuse' proforma to record findings.[9] If immediate specialist review has been obtained, some parts of this examination can be left to the referral unit so the child does not need to have two lengthy examinations. Specialist units will also use forensic photography to document injuries.

ANSWER 6

Several explanations could account for markings shown in *Figure 2*. One possibility is abuse. Another is that these markings represent a self-inflicted 'baby hicky'. For example, when a baby's dummy falls on the floor, a baby may suck furiously on their forearm in the location of the bruises. This behaviour may be witnessed by parents and/or healthcare providers (GPs, practice nurses) and is one cause of non-assaultive bruising in babies. Breastfed babies have also been reported to have self-inflicted bruises on their arms from sucking when hungry.[6]

As a general principle, clinicians need to assess a baby's stage of development when deciding whether bruises could be accidental. This is more important than age. If a baby is cruising or walking, this activity has a direct bearing on the amount and location of non-abusive bruising that the child may have.[10] Once children are cruising, bruising becomes more common from falls and, in active childhood, children sustain more bruises.[11]

ANSWER 7

Common sites of accidental childhood bruising are described below:[11]

- In mobile children, the most common sites of bruising are the shins and knees.
- Most accidental bruises occur over bony prominences and are commonly seen on the front of the body. These correspond to the sites that are bumped in falls.
- In slips, trips and falls, the most common sites of bruising are the back of the head and the front of the face, including the T zone of the forehead, nose, upper lip and chin.
- Children who are pulling to stand may bump their head and sustain bruising to the head, usually on the forehead.

ANSWER 8

Accidental bruising in childhood is uncommon in a number of sites, including the back, buttocks, forearm, face, neck, ears, behind the ears, abdomen or hip, upper arm, posterior leg, foot and/or hands.[12] Bruising in any of these areas should be treated seriously by healthcare professionals and requires questioning, carefully recording any explanation and, where there is doubt, reporting and further investigation. If practicable, speak to the child alone and ask them for an explanation of the bruises before asking adults. Cast your eyes over all children present and look for signs of bruising.

CONCLUSION

It is important to treat Kylie with kindness and respect as you explain the need for urgent assessment and mandatory reporting. It is generally helpful to emphasise the need for immediate medical assessment rather than to emphasise the child protection concerns. This minimises distress for the parent if there is an innocent explanation and makes any police investigation easier, as the actual perpetrator is less likely to be alerted to prepare a cover story, which may, for example, involve coercing older children to say that they 'dropped the baby', etc. The family GP will be providing a professional response, rapid referral and immediate reporting. In a rural situation, where there may be delay, a clinical photograph of the injury, included in your medical record, may assist in preserving information for specialist child protection review. Your records, including any photograph could have legal implications and any details obtained of the history and examination, would be recorded with special care.

Abuse and violence: Working with our patients in general practice[5] (the White book), provides excellent coverage of these issues in the chapter 'Child abuse'. There is useful information and downloadable body diagrams on The Royal Children's Hospital, Melbourne Web site (www.rch.org.au/clinicalguide/guideline_index/Child_Abuse/).

All GPs are involved in primary prevention of child abuse by supporting healthy families and parenting, but secondary prevention in terms of detecting abuse is also vital.

This case highlights the potential serious significance of any bruising in a pre-ambulant baby. Infants cannot verbally disclose what is happening or take evasive action and, given their fragility, are at particular risk of death or serious injury from physical assault.

RESOURCES FOR DOCTORS

- The Royal Australian College of General Practitioners' White book has a range of resources by state and territory to support the management and mandatory reporting of child abuse, visit www.racgp.org.au/your-practice/guidelines/whitebook/tools-and-resources/7-resources
- The Royal Children's Hospital Melbourne, www.rch.org.au/clinicalguide/guideline_index/Child_Abuse/

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CASE 6

JOHN IS STRUGGLING AT SCHOOL

John, 8 years of age, is a year 3 student who is brought in by his mother, Susan. She reports that he is struggling with reading and writing at school. Susan believes this has been present since John started school, but it seems to have worsened. She believes this is affecting his learning ability.

When John reads, he often skips words, misreads words and lacks accuracy in reading words. As a result, he is reluctant to participate in reading activities.

John also has problems with spelling words and handwriting skills.

QUESTION 1

What history would you take from John's mother?

QUESTION 2

What physical examination would you perform on John?

QUESTION 3

What collateral information would assist with your assessment?

FURTHER INFORMATION

Susan recalls that John had an unremarkable antenatal history and was born at full term without complication. He is the eldest of three children. He started talking at the age of 2 years and a speech pathologist diagnosed him with a moderate language delay at the age of 3 years. At that time, he often cried, as he could not always communicate what he wanted. Susan attended a speech program course that involved using simple sign language to communicate, sound repetition and simplified sentence structure.

When John was 5 years, Susan noticed he was unable to read or write as fluently as other children. He had no trouble with speaking, constructing sentences or pronouncing words. His behaviour at school and home were consistent with a good attention span. Recently, John has become frustrated with falling behind in his academic work, which has resulted in occasional teasing at school and irritability at home. There have been no issues with disorganisation, self-care, eating or sleeping habits.

John's physical examinations are unremarkable and there are no dysmorphic features. He is interactive throughout the consultation. His weight is 25 kg and height is 130 cm (50th centile). His reading ability is consistent with prep level and he has difficulties with spelling simple words and constructing simple grammatical sentences. He has a good attention span and is focused throughout the activity. His conversation content is appropriate for his age. He has a particular interest in astronomy and science.

QUESTION 4

What is your diagnosis?

QUESTION 5

What are other possible causes to consider in a child who struggles at school?

QUESTION 6

What referrals might be appropriate in a case such as John's?

QUESTION 7

How would you manage and support John in the community?

QUESTION 8

What is the long-term prognosis for John?

CASE 6 ANSWERS

ANSWER 1

The approach to assess a child who is struggling at school is to determine if there are any biological and environmental risk factors, rule out any organic medical conditions and work out whether there is an appropriate diagnosis.[1]

First, assess the duration of his school difficulties. Any recent deterioration in school performance may be caused by family stressors and/or psychological stressors. It is important to determine whether it has been a chronic issue, especially with early milestone delays (social history from birth).

In a clinical situation, it is best to speak to the child directly (depending on the age of the child).[1] Ask the parents and child how they know if they are struggling at school. You can gather information by asking the following types of questions (collateral history from teachers):

- Have you been told by a teacher?
- What is the specific area of difficulty (eg reading, writing, mathematics, poor attention span, difficulty completing a given task, difficulty retaining memory and/or facts)
- Which subjects does the child enjoy and why?
- What is the child good at?
- Does the child have any particular hobbies/interests?
- How do they get on with teachers and peers in school?

You need to determine whether the problem is a specific learning disorder (literacy and numeracy) or a more pervasive disorder, such as attention deficit hyperactivity disorder (ADHD).[1]

It is important to explore the child's social and home life. This can be done by:

- Exploring the nature of John's friendships at school.
- Determining if there is any bullying.
- Asking if parents have discussed the concerns with teachers/principal, whether the school has taken any actions and, if so, what were the outcomes.
- Exploring John's home environment, including family composition and sibling orders.
- Asking how he gets along with his siblings and if he has any behavioural issues at home.
- Asking about his degree of organisation at home, such as organising himself for school and the level of supervision required for doing homework.
- Determining how much screen time he is exposed to.[1]

You should take a thorough maternal pregnancy history including exposure in utero to drugs/alcohols and maternal depression. It is also important to take a perinatal history to consider prematurity and low birth weight.[1] Explore any past history of developmental or behavioural concerns, such as speech/language delay, hearing problems and difficulties with social interaction. Ask for any previous history of settling issues and sleeping/feeding difficulties as a baby.

Document the child's relevant past medical history such as immunisation, sleep history, dietary history, any chronic illness or prior significant medical illness. Exclude any history of neurological problems.

Family history is important, especially if John has first- or second-degree relatives with literacy difficulties at school or speech delays.[1]

The Royal Children's Hospital's guidelines[2] for assessment are shown in *Table 1*.

Table 1. Initial work-up assessment[2]

- Onset and course of symptoms
- Family history of similar patterns
- Hearing and vision assessment
- School history
- Contributing causes (eg anxiety, family dysfunction, auditory processing problems)
- Comorbidities (eg ADHD, other behaviour disorders, language disorders, developmental disorders, intellectual disability)
- PEDS Screening Tool (parents' evaluation of developmental status)

ANSWER 2

During physical examination, you should measure John's weight, height and head circumference. Check for any dysmorphic features and birthmarks that may suggest an underlying neurocutaneous disorder.[1]

Conduct a general system examination to assess:

- visual acuity
- strabismus
- ear, nose, throat
- muscle tone, muscle strength and coordination.

You can ask John to perform simple tasks to assess his fine motor skills. This could include undoing buttons or zippers, tying shoe laces, recognising left and right, and assessing writing skills, sentence construction and grammar skills.

During the assessment, you should observe John's social interaction and speech to determine if there is a receptive or expressive language delay. Also observe his ability to answer questions directly with complex sentences and his ability to hold an interactive conversation. Listen carefully for any mispronunciation of words.

You could also ask John to read an age-appropriate book to assess how accurately and fluently he can read. You can check John's comprehension by asking him questions from the book.

ANSWER 3

You could ask John's parents to provide any past and recent school reports to review his academic grades and the comments made by teachers. You could offer to speak directly to John's teacher regarding his school progress and what strategies/interventions have been tried to assist John. This may be done during an organised telephone consultation with the teacher in the presence of the parents. The child could also be present. Alternatively, you could ask for written reports from the teacher and/or give the teachers the option to contact you.

If the school has an educational psychologist, the GP or parents could request special assessment by the psychologist in class to identify areas of the child's weakness.

ANSWER 4

John has a specific learning disorder (SLD) in reading and writing (ie dyslexia).

According to the Diagnostic and Statistical Manual of Mental Disorders, fifth edition (DSM-5),[3,4] SLD is a neurodevelopmental disorder. This means an impaired ability to read, write and perform mathematics. SLD is a specific clinical diagnosis only. Not all children with a learning disability identified by the school meet the criteria of SLD.

The DSM-5 lists four diagnostic criteria to be met for a diagnosis of SLD. These criteria are summarised below.[3,4]

- Learning difficulties and impaired academic skills have persisted for ≥6 months despite extra assistance.
- The measured clinical assessment of academic skills is significantly below the chronological age of the patient (using standardised individual assessment) and this causes impairment in everyday living activities)
- Symptoms manifest in school-age children but can sometimes manifest in adults.
- Intellectual disability, visual/hearing impairment, mental/neurological disorders and psychosocial factors must be ruled out.

ANSWER 5

There is a range of other causes of learning difficulties:

- Biological and genetic factors should be considered and it is necessary to ask about the family history of genetic disorders or chromosomal abnormalities.[1]
- Organic medical conditions: ask about any significant chronic illness, epilepsy (especially absence seizures), neurological problems, poor sleeping quality, obstructive sleep apnoea, iron deficiency, congenital abnormalities (eg Kleinfelter's syndrome).[1]
- Social issues/psychological stressors: lack of parental stimulation, parental neglect, child abuse, multiple relocations, low socioeconomic class, parental conflicts, excessive screen time, lack of support at home.[1]
- School issues: ineffective school system, lack of exposure to academic work, English as a second language, school bullying.[1]
- Mental health: depression, anxiety, social anxiety disorder, panic attack, specific/general phobia, post-traumatic stress disorder (PTSD), acute stress event, grieving, drugs/alcohol issues, oppositional defiance disorder, conduct disorder, parasomnia.[1]
- Pervasive disorder: ADHD.[1]
- Underlying intellectual disability and autism spectrum disorder.[1]
- Vision/hearing impairment.[1]

Note that all of these symptoms and conditions can co-exist with a specific learning disorder.

ANSWER 6

John should be referred to a paediatric audiologist and a paediatric ophthalmologist for a formal hearing and vision assessment, respectively. This should be done for any child who is struggling at school.

Encourage John's parents to discuss his needs with his school and attend special learning support meetings with teachers. John's parents should be encouraged to find out what the school is able to offer and how often the school is able to provide help (eg individual assistance in a small group). The head of special education services at school can assess and plan alternative learning strategies/styles and offer catch-up individual sessions. The special education program is dedicated to support the educational needs of students with one or more disabilities, including:

- autism spectrum disorder
- hearing impairment
- intellectual disability
- physical impairment
- speech-language impairment
- vision impairment.

Referral for psycho-educational assessment by an educational or developmental psychologist is useful for assessing the extent of any learning difficulty and to exclude any underlying intellectual disability. This assessment usually involves testing intellectual functioning (IQ test) and academic ability, and assessing a child's attention and behaviour.

Other aspects of assessment include visual and verbal problem solving skills as well as auditory processing of information and memory processing. AUSPELD provides a list of appropriate psychologists (refer to *Resources*).

A psychologist will use a parent questionnaire as part of the assessment (eg the Autism Spectrum Rating Scales (ASRS), the Connors questionnaire for ADHD or the PEDS screening tools. The psychologist will provide specific parent training around managing behavioural and attention issues.

A referral to a speech therapist is worthwhile, even if John's speech and language seem adequate. A speech therapist is available privately, through public hospitals, speech pathologist visits to schools, or through community child health clinics. Speech therapy is effective for expressive phonology, expressive vocabulary and expressive syntax difficulties. It is less effective for treating receptive language disorders.[1]

Referral to an occupational therapist will be needed to assess John's fine motor skills and handwriting skills. This type of assessment is beneficial for children who have difficulty with written language.

Referral to a paediatrician is recommended if there are concerns about attention, organisation, behaviour and social skills, or if there were an underlying complex medical cause.

Assessment by a child psychiatrist may be necessary if there were complex behavioural and mental health concerns.

If available, referral to a multidisciplinary learning difficulty clinic should be considered.

External tutors could be used, tailoring to the child's strengths and weaknesses. You can recommend specific training in phonics, sightwords and decoding, which is beneficial for children with reading and spelling difficulties.[1]

You could recommend that John use the software spellchecker or ask others to read through his written work.

The Royal Children's Hospital's guidelines[2] recommend referral if:

- the child not functioning as expected in school
- the cause of the learning problems not clear
- routine school supports not effective or not sustained
- previous assessments are not well understood or integrated into school or homework programs
- the child develops anxiety or has low self-esteem
- there is significant parental concern (eg evident from the PEDS screening tool).

ANSWER 7

The GP has an important role in overseeing John's overall progress. It is important to support the whole family throughout John's school years, focusing on John's strengths and weakness in order to develop a comprehensive multidisciplinary approach.

As a GP, you can empower John and his family to ensure that community resources are used effectively. The GP's role includes:

- early referral to an educational psychologist or other appropriate allied health professional
- early referral for hearing and vision assessment
- follow-up of results
- providing clear communication and education for the parents
- regular monitoring of the child's progress
- direction/guidance on any changes in the level of input from different specialties, as needed.

ANSWER 8

John has a specific reading and writing disorder, which is a lifelong condition. These children are at a higher risk of problems with long-term academic and behavioural issues. [1]

John's parents need to be informed that this does not mean he will be an unsuccessful adult or have a poor quality of life. The parents should be reassured that John can still learn, but a different approach will be needed. Extra time should be given to assess John's knowledge.

John's long-term success will depend on the amount of support he receives from his family, school and future employers. Creating a specific management plan tailored to John's strength and weakness will also contribute to his future success. [1]

Choose a university that can accommodate students with learning difficulties, for example, by allowing extra time in exams, using laptops, and text-to-speech programs.

Other options for those who struggle at school would be a combined school program, work experience and vocational training. This provides extra opportunities of training to equip students for future employment.

CONCLUSION

For children with specific literacy difficulties (specifically, reading and writing), early identification and intervention is crucial. Reading and writing skills determine a child's academic success, future work success, self-esteem and confidence. [1]

Reading and writing is fundamental in determining the level of education a child may eventually achieve and ultimate future success.

It is important that GPs assess children holistically and not overlook assessing a child's literacy and numeracy skills at school. Early identification allows for early assessment and referrals to allied health professionals. The appropriate systematic phonic-based program can be started early to maximise benefits in reading and writing.

It is important that the GP, being the first point-of-contact for patients, continue to monitor the child's overall progress in conjunction with specialists and allied health professionals. Simple and effective assessment methods can also be used in the consultation at regular intervals to monitor a child's progress.

RESOURCES FOR DOCTORS AND PATIENTS

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- AUSPELD supports people with learning disabilities. Its website lists the state-based SPELD associations, http://auspeld.org.au
- Dyslexia Australia provides assessments, evaluations and solutions, www.dyslexia-australia.com.au
- The Department of Education, Training and Employment has a range of special education programs, http://education.qld.gov.au/studentservices/learning/disability/parentguide/programs-and-services/programs
- Speech Pathology Australia, www.speechpathologyaustralia.org.au

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ACTIVITY ID: 21109

STAGES OF LIFE: CHILDHOOD

This unit of *check* is approved for 6 Category 2 points in the RACGP QI&CPD program. The expected time to complete this activity is 3 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 *gplearning* website, http://gplearning.racgp.org.au
- answering the following multiple choice questions (MCQs) by logging on to the *gplearning* website, http://gplearning.racgp.org.au
- you must score ≥80% before you can mark the activity as 'Complete'
- completing the online evaluation form.

You can only qualify for QI&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.

QUESTION 1

Jasper is 4 years of age and has had a 1-month history of coughing during the day and night, and episodes of wheezing about 3 nights a week. History taking and physical examination lead you to a diagnosis of asthma.

According to the National Asthma Council Australia definition, how would you classify Jasper's symptoms?

A. Mild persistent asthma

B. Moderate persistent asthma

C. Severe persistent asthma

D. Frequent intermittent asthma

QUESTION 2

How would you manage Jasper's asthma?

A. Commence treatment with a low dose of an inhaled corticosteroid as a preventer plus a β2-receptor agonist as a reliever and review Jasper in 4 weeks.

B. Commence treatment with an inhaled corticosteroid and magnesium supplementation and review Jasper in 2–3 months.

C. Commence treatment with an inhaled corticosteroid and montelukast.

D. Recommend lifestyle modifications to remove dust mite exposure.

QUESTION 3

James is 6 years of age and has tested positive for coeliac serology. He has come to see you today with his mother to discuss his management. What is the most appropriate next step?

A. He should commence a gluten-free diet.

B. He should commence a lactose-free diet.

C. He should be referred for a duodenal biopsy.

D. He should be referred to a paediatric dietitian.

QUESTION 4

James's mother is planning a second pregnancy and asks if there is anything she can do to prevent coeliac disease in her new baby. Which of the following statements is correct?

A. A gluten-free diet during the pregnancy will not alter the risk of the baby developing coeliac disease.

B. Research has shown that breastfeeding reduces the risk of developing coeliac disease.

C. Gluten should be excluded from the baby's diet if the baby develops symptoms of coeliac disease.

D. Gluten should be excluded from the baby's diet for the first 6 months.

QUESTION 5

Sally, 5 years of age, has had episodes of distressing abdominal pain since starting school 2 months ago. Which of the following additional features, if present, would support a diagnosis of idiopathic constipation?

A. Decreased bowel sounds

B. Weight loss

C. Vomiting

D. Encopresis

QUESTION 6

Lizzie is concerned about her baby, Timmy, and brings him to see you. Timmy is 6 weeks of age and for the past 2 weeks he has been crying more than usual, seems unsettled and irritable, and vomits occasionally. Timmy is being breastfed and has a feed every 2–3 hours.

What element of Timmy's history would exclude cow's milk protein allergy (CMPA) as a cause of his symptoms?

A. Timmy is being breastfed.

B. Timmy needs to be fed every 2–3 hours.

C. Timmy seems unsettled and irritable.

D. Timmy's age.

QUESTION 7

Which of the following is the best management option for a baby with uncomplicated reflux?

A. Introducing thickening feeds
B. Treatment with a prokinetic drug such as metoclopramide
C. Providing the parents with patient education, including feeding modifications and avoiding exposure to tobacco smoke, and monitoring the baby's growth
D. Treatment with a proton-pump inhibitor (PPI).

QUESTION 8

Which of the following sites of bruising in a mobile child should raise suspicion of abuse?

A. Shins and knees
B. Bony prominences on the front of the body
C. Back of the head
D. Forearm

QUESTION 9

What immediate course of action should you take for a pre-ambulant baby who presents with bruising on the arms and legs?

A. Contact the child protection helpline for your state.
B. Admit the baby to hospital for further assessment and to restrict parental contact until the cause of bruising has been identified.
C. Report the bruising to the police.
D. Assess whether the baby is being abused.

QUESTION 10

Which of the following is one of the Diagnostic and Statistical Manual of Mental Disorders, fifth edition (DSM-5), diagnostic criteria for specific learning disorders (SLDs)?

A. Learning difficulties have persisted for 3–4 months.
B. Assessment by the teachers indicates inadequate academic performance.
C. Symptoms manifest in school-age children but can also manifest in adults.
D. Symptoms are associated with intellectual disability.

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