Common neonatal presentations to the primary care physician

Alicia Quach

This article is the first in a series on paediatric health. Articles in this series aim to provide information about diagnosis and management of presentations in infants, toddlers and pre-schoolers in general practice.

Background
Newborn babies are very vulnerable in their first weeks of life. Timely and appropriate management of neonatal conditions is paramount for health and developmental outcomes.

Objectives
The aim of this article is to provide an overview of common neonatal presentations to general practice, and highlight significant conditions that may require referral to the emergency department and/or other specialist.

Discussion
Clinical history and examination are the most important tools in neonatal assessment. Babies often present with non-specific symptoms, but a thorough clinical assessment can identify the ‘unwell baby’ who requires immediate transfer to hospital. This includes babies with sepsis, moderate-to-severe dehydration or who are in acute cardiorespiratory compromise. A comprehensive neonatal assessment will also help to differentiate babies with significant conditions that may warrant further specialist input from those with normal neonatal development where parental support and reassurance may be sufficient.

Fever
Fever (rectal temperature >38°C)²,³ in a newborn baby can be the first indicator of a serious invasive infective illness. Conversely, hypothermia (rectal temperature <36.5°C)⁴ can also be a sign of sepsis, as neonates have difficulty regulating temperatures.⁵ The health of neonates with sepsis can deteriorate rapidly; therefore, if a fever is detected (gold standard is to take the rectal temperature, but axillary or temporal artery temperature are acceptable), the baby requires immediate referral to the emergency department for a full septic workup, and hospital admission for empirical antibiotics. Ambulance retrieval may be necessary for babies who have associated signs of haemodynamic instability, acute respiratory distress and/or are non-responsive. A full septic workup should include a full blood count and film, blood culture, urine culture (through aseptic suprapubic aspiration), lumbar puncture and, if clinically indicated, a chest X-ray.¹ Where possible, it is recommended that neonates with suspected sepsis be admitted to hospital and treated with intravenous antibiotics; oral antibiotics may lead to only partial treatment and false-negative culture results.⁶

Respiratory symptoms
Respiratory symptoms are common and the majority will be benign. However, it is important not to miss the acutely unwell baby in respiratory distress, as these babies should be transferred to the emergency department via ambulance.⁷ Table 2 outlines the signs and causes of acute respiratory distress outside the first 24 hours of life, and other common respiratory presenting concerns and conditions. In neonates, a cough can be due to a common viral upper respiratory tract infection, but it can also be a sign of a more significant pathology. Babies who have a cough and any associated ‘red flag’ signs or symptoms outlined in Table 2 should be referred for further investigation with a paediatrician. Clinical suspicion of pertussis infection warrants referral to the emergency department for laboratory confirmation, antimicrobials and monitoring, as these babies are at greatest risk of complications with apnoea, pneumonia, encephalopathy and death.⁸

Neonates are obligatory nose breathers, and nasal congestion with mucus often results in noisy breathing. Normal saline drops or spray may relieve some of the nasal congestion. The most common pathological cause of noisy breathing is laryngomalacia. This developmental anomaly causes stridor through collapse of the supraglottic structures during inspiration. Babies with mild laryngomalacia who are feeding well and thriving can be regularly reviewed in the GP setting. Parents should be advised that stridor may become louder in the first six
months of life, but will usually resolve by 12–18 months. Babies who have associated complications (eg poor feeding, gastro-oesophageal reflux [GOR]) should be referred to a respiratory paediatrician or otolaryngologist for further assessment.9

Parental concerns regarding irregular breathing or pauses in their baby’s breathing are also common GP presentations. In the majority of neonates, these irregularities will be due to ‘periodic breathing’, which is a normal developmental phenomenon. Periodic breathing is characterised by alternating cycles of five to 10 seconds of breathing and pauses in breathing. It is not associated with bradycardia or cyanosis. It increases in frequency between two and four weeks of age and resolves by six months of age.10

Apnoea is defined as pauses in breathing of greater than 20 seconds, or shorter duration if accompanied by cyanosis or bradycardia.10,11 This is of great concern, and a significant medical cause needs to be excluded. If a medical cause is not evident following clinical assessment, these babies are classified as having had a brief resolved unexplained event (BRUE). BRUE replaces the previous terminology: apparent life threatening event (ALTE).10 BRUEs can be stratified into low risk and high risk, where low-risk BRUEs generally do not require hospital admission or invasive testing.12 All neonatal BRUEs are categorised as high risk, given the age of the baby,12 and should be reviewed by a paediatrician for further investigation. Table 2 summarises common causes of apnoea.

### Gastrointestinal symptoms

Small amounts of effortless posseting or physiological GOR are common in babies. In otherwise well babies who are feeding adequately and thriving, parental reassurance that this is most likely to improve in the first year of life is sufficient. General measures, such as holding the baby in the prone position after feeds and thickening agents, may help reduce the vomiting. Acid-suppression agents (ie H2-agonists, proton-pump inhibitors) should be reserved for babies with associated complications, such as inadequate weight

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**Table 1. General principles for a routine neonatal assessment**

<table>
<thead>
<tr>
<th>History</th>
<th>Examination</th>
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<tbody>
<tr>
<td>Maternal or antenatal history</td>
<td>• Observe interaction with carers</td>
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<tr>
<td>• Relevant maternal medical history</td>
<td>• Alertness of baby</td>
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<tr>
<td>• Antenatal visits</td>
<td>• General colour and tone</td>
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<td>• Investigations during pregnancy</td>
<td>• Signs of dysmorphism</td>
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<td>• Fetal growth</td>
<td>• Inspect for skin lesions throughout examination</td>
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<td></td>
<td>• Weight, length, head circumference measurements</td>
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<td></td>
<td>• Systematic head-to-toe examination</td>
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<td></td>
<td><strong>Head and neck</strong></td>
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<td>• Fontanelle</td>
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<td>• Sutures</td>
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<td></td>
<td>• Oral cavity (eg palate)</td>
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<td>• Ears (eg position, pits)</td>
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<td></td>
<td>• Neck (eg masses, range of movement)</td>
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<td>• Red eye reflex can be left to end of examination</td>
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<td></td>
<td><strong>Chest</strong></td>
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<td>• Cardiac examination (eg heart rate)</td>
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<td>• Respiratory examination (eg respiratory rate)</td>
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<td>• Chest deformities</td>
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<td><strong>Abdomen</strong></td>
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<td>• Umbilicus (eg hernia, granuloma, infection)</td>
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<td>• Palpate for organomegaly, hernias</td>
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<td>• Femoral pulses</td>
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<td><strong>Genitals</strong></td>
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<td>• Patent anus</td>
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<td>• External genitalia</td>
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<td>• Position of testes in male</td>
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<td>• Position of urethral meatus</td>
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<td><strong>Limbs</strong></td>
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<td>• Digits</td>
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<td>• Symmetrical movements</td>
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<td></td>
<td><strong>Hips</strong></td>
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<td></td>
<td>• Barlow and Ortolani manouevres</td>
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<td></td>
<td><strong>Back</strong></td>
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<td></td>
<td>• Ventral suspension</td>
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<td></td>
<td>• Spinal dysraphism</td>
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<td><strong>Reflexes</strong></td>
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<td>• Rooting</td>
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<td>• Tonic neck or fencing</td>
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<td>• Grasp</td>
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<td>• Stepping</td>
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Peripartum history
- Gestational age
- Delivery mode
- Resuscitation, Apgar scores
- Vitamin K given
- Birth weight
- Nursery or intensive care admission
- Interventions (eg oxygen, nasogastric feeds, phototherapy, antibiotics)

Postnatal period
- Feeding method
- Sleep or settling patterns
- Wet or dirty nappies
- Growth
- Community maternal nurse visits
- Social context – supports, maternal mental and physical health, is family coping?

Address any concerns
# Table 2. Differential diagnoses for respiratory symptoms and signs

<table>
<thead>
<tr>
<th>Clinical presentation</th>
<th>Differential diagnoses</th>
<th>Red flags or supporting features</th>
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| **Acute respiratory distress** (eg tachypnoea, accessory muscle use, central cyanosis, nasal flaring, expiratory grunting) | Infection | • Fever or hypothermia  
• Irritability or lethargy  
• Decreased feeds or poor urine output  
• Infectious contacts |
| Foreign body | | • Acute onset  
• Associated stridor or wheeze |
| Trauma | | • Physical signs of trauma (eg bruising)  
• Suspicion of non-accidental injury  
• Seizures |
| Congenital heart disease | | • Cyanosis  
• Cardiac murmur  
• Failure to thrive |
| Metabolic acidosis | | • Large volume fluid losses (eg vomiting, diarrhoea)  
• Failure to thrive  
• Apnoea  
• Seizures |
| **Cough** | Respiratory infection | • Coryzal symptoms  
• Infectious contacts  
• Prolonged episodic coughing (red flag for *Bordetella pertussis*)  
• Fever |
| Tracheo-oesophageal fistula | | • Coughing and choking with feeds  
• Antenatal polyhydramnios |
| Chronic lung disease | | • Prematurity  
• Prolonged intubation |
| Tracheo-bronchomalacia | | • Cough present since birth  
• Barking cough |
| Congenital heart disease | | • Cough with cyanosis  
• Cardiac murmur  
• Failure to thrive |
| **Noisy breathing** | Laryngomalacia | • Stridor (ie noisy breathing on inspiration) worse in supine position |
| Tracheomalacia | | • Noisy breathing on expiration  
• Barking cough |
| Laryngeal / subglottic mass | | • Cutaneous lesion (eg haemangioma) over face, neck or upper chest region |
| Choanal atresia | | • Grunting noise  
• Cyanosis with feeding (if bilateral)  
• Unilateral nasal discharge |
| Vocal cord paralysis | | • Hoarse cry  
• Other midline deformities |
| **Apnoea** | brief resolved unexplained event | • Apnoea with colour change, change in muscle tone, altered conscious state that completely resolves within one minute  
• Other medical causes excluded on clinical assessment |
| Apnoea of prematurity | | • Baby <37 weeks’ gestation  
• History of oxygen support |
| Infection | | • Refer to acute respiratory distress |
| Head trauma | | • History of birth asphyxia requiring resuscitation  
• Risk factors for abusive trauma |
| Structural airway obstruction | | • Facial dysmorphic features  
• Congenital malformations in chest or abdomen |
gain, oesophagitis or aspiration. There is no clear causal link between GOR and infant irritability, and anti-reflux medication is generally not warranted in these instances. Vomiting as a result of a more serious condition, such as pyloric stenosis, intestinal obstruction, sepsis or neurological cause (eg subdural or intracranial haemorrhage, hydrocephalus), needs to be promptly referred to the emergency department. Red flags for these conditions include projectile vomiting immediately post-feeds (associated with demands to be re-fed soon after), bilious vomiting, acute abdominal distension, fever, lethargy, dehydration or bulging fontanelle.

There is no universally agreed clinical definition of constipation for neonates. They may pass bowel motions several times a day or have more than a week between bowel motions. Formula-fed babies typically produce firmer and less frequent stools than breastfed babies, but unless these are hard and pellet-like, the baby is unlikely to be constipated. Some babies will strain and cry for longer than 10 minutes before passing soft stools. This phenomenon, known as dyschezia, is caused by an inability to coordinate the increase in intra-abdominal pressure with pelvic floor relaxation. It is a functional, self-limiting condition, and is not due to constipation. Caution should be applied and organic pathology excluded before prescribing laxatives in neonates. Clinical history and examination will detect some significant conditions, including:

- Hirschsprung’s disease, alerted by a history of delayed meconium passage (after 48 hours of life)
- mechanical bowel obstruction suspected with firm abdominal distension on palpation
- spinal dysraphism leading to autonomic or sphincteric dysfunction.

Cutaneous lesions over the sacrococcygeal region may be indicative of closed spinal dysraphism.

It is also normal for newborn babies to have frequent, loose stools. Babies who have true diarrhoea will produce more watery and more frequent stools than usual. The most common causes of diarrhoea in neonates are viral or bacterial infections or cow’s milk protein allergy (CMPA). Babies with acute infective diarrhoea (gastroenteritis) need to be monitored closely for dehydration. Table 3 outlines the signs of dehydration and other signs of the unwell baby that should prompt early transfer to hospital. Admission to hospital should also be considered for those who are unlikely to maintain adequate oral intake at home. Antimicrobial therapy is rarely warranted in gastroenteritis, as most cases are viral and/or self-limiting.

Unsettled baby

All newborn babies cry. Normal infant crying patterns tend to increase in duration each week by week, peaking at around six to eight weeks of age, and receding to lower, stable levels at around four to five months of age. The typical presentation is clustered periods of inconsolable crying, some for more than two to three hours in duration, often in the late afternoon and evening. In otherwise well babies, reassurance, support and review in the first few months can be therapeutic tools in their own right. The exclusion of pain or ‘wind’ as the cause of crying will help to reassure parents. Box 1 lists some parental education resources on normal infant development, unsettled babies and breastfeeding. Alternative therapies, such as simethicone, herbal treatment, acupuncture and manipulation techniques, are not supported by the evidence. Advice to change from breastmilk to formula, or to change between formula brands, should be avoided. There is emerging evidence that probiotics may be helpful in settling breastfed babies, but currently there is no universal consensus for this to be a standard recommendation.

As part of the assessment of an unsettled baby, it is important not to miss a pathological cause. If there is a sudden onset of persistent crying, an acute pathology such as infection, hair tourniquet (ie strangulation of an appendage or digit by a thread-like material, such as hair), corneal abrasion and non-accidental injury should be considered. CMPA is a recognised cause for the unsettled baby. Supporting features for CMPA include blood and mucus in the stool, diarrhoea or constipation, inadequate weight gain, eczema, and family history of atopy.

A cow’s milk exclusion diet (including mothers if breastfeeding) may be trialled in these babies to confirm the diagnosis. In babies who are formula-fed, a trial of extensively hydrolysed formula and/or amino acid formula will be required. Soy infant formulas are not recommended in infants younger than six months of age. Rice protein-based formulas can be used as a short-term, non-prescription alternative while awaiting specialist review. Referral to a paediatrician or allergy specialist and dietitian is recommended for suspected cases of CMPA to ensure adequate parental education and future dietary management.

Feeding difficulties

Prematurity (gestation <37 weeks) is the most common cause of feeding difficulties in neonates. Their immature physiology can result in discoordination between sucking, swallowing and breathing. Breastfeeding, however, can be a challenging process for any new mother. Support from a maternal child health nurse or lactation consultant can result in positive outcomes for both mothers and their babies. Tongue-tie, or ankyloglossia, has long been linked with difficult breastfeeding and maternal nipple pain. A Cochrane meta-analysis found that frenotomy (surgical release of tongue-tie) reduced maternal nipple pain in the short term, but did not find consistent positive effects on breastfeeding. Maternal nipple pain is, however, a common reason for cessation of breastfeeding and early referral for frenotomy may prevent this.

Breastfeeding may not be a viable option for all mothers, and advice regarding infant formula, expressed breast milk, or a mixture of the two should be made available in a supportive and non-judgemental manner. Babies can lose up to 10% of their birthweight in the first week of life, and may take a further two weeks to regain their birthweight.
It is therefore more important to track the actual weight difference in grams between visits. Following the initial postpartum weight loss, newborns are expected to gain 30–40 grams per day on average. Failure to thrive or inadequate weight gain is most commonly a result of inadequate oral intake. If a baby continues to display inadequate weight gain despite increased feed frequency and/or supplementary feeds, referral to a paediatrician for further investigation and management should be made.

**Jaundice**

Jaundice, or hyperbilirubinaemia, is the result of bilirubin pigment deposition in the skin and mucous membranes. In the majority of cases, jaundice in neonates is due to unconjugated hyperbilirubinaemia. Conjugated hyperbilirubinaemia or ‘cholestatic jaundice’ is always pathological, and detection of this should prompt immediate review with a paediatric gastroenterologist. The following discussion is in relation to unconjugated hyperbilirubinaemia.

Visual assessment of jaundice alone is an unreliable indicator of the degree of hyperbilirubinaemia. Assessment should include detection of any signs of bilirubin toxicity (Table 3), and identification of risk factors, including the following:

- Prematurity
- Jaundice within the first 24 hours
- Blood group incompatibility
- Cephalohaematoma or other birth-related trauma
- Weight loss >10% of birthweight
- Previous sibling with hyperbilirubinaemia requiring treatment.

Babies with jaundice and added signs of bilirubin toxicity require immediate referral to hospital. Babies with prolonged jaundice who are otherwise well looking, feeding adequately and with no risk factors are most likely to have physiological jaundice or breastmilk jaundice, and can be managed as outpatients. A bilirubin level (total and fractionated) should be checked with early follow-up for results and clinical review. Bilirubin threshold tables (www.nice.org.uk/guidance/cg98) should be used to determine whether the baby requires treatment with phototherapy or exchange transfusion.

**Rashes**

In any baby who presents with a vesiculopustular rash, significant causes such as bacterial, viral and fungal infections need to be considered. If a rash is accompanied by systemic signs of being unwell, such as fever, lethargy or poor feeding, then the baby needs to be referred immediately to the emergency department for further assessment. Recent exposure to infectious diseases such as Varicella-Zoster virus (VZV) or Herpes simplex virus (HSV), should also alert the physician to the possibility of an invasive infective disease.

Common benign rashes that may present in the newborn include erythema toxicum and milia. Erythema toxicum

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**Table 3. Signs and symptoms of an unwell baby**

<table>
<thead>
<tr>
<th>Vital signs</th>
<th>Heart rate (bradycardia &lt;110 beats/minute and tachycardia &gt;170 beats/minute)</th>
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<tbody>
<tr>
<td></td>
<td>Temperature (fever &gt;38°C; hypothermia &lt;36.5°C)</td>
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<tr>
<td></td>
<td>Respiratory rate (bradypnoea &lt;25 breaths/minute and tachypnoea &gt;60 breaths/minute)</td>
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**Signs of dehydration**

- Weight loss (bare) in setting of acute illness or >10% of birthweight
- Decreased urine output
- Dry mucous membranes
- Sluggish capillary refill (>2 seconds)
- Poor tissue turgor
- Sunken eyes and anterior fontanelle

**Systemic specific signs**

- Acute respiratory distress: tachyphoea, accessory muscle use, grunting, nasal flaring, central cyanosis
- Gastrointestinal: acute abdominal distension that is firm, bilious vomiting, projectile vomiting
- Cardiac: cyanosis, cardiac murmurs
- Severe jaundice with signs of bilirubin toxicity: lethargy, dehydration, pallor, irritability, hypotonia or hypertonia, seizures, fever

**Non-specific signs**

- Lethargy
- Poor feeding
- Inadequate weight gain
- Irritability – persistent
- Rashes
- Seizures
is a benign, self-limiting skin condition categorised by small erythematous papules, vesicles and pustules. It affects 30–70% of newborns, typically within the first two weeks of birth. Erythema toxicum can be differentiated from an infective rash by its tendency to wax and wane over several days. It is also unusual for an individual erythema toxicum lesion to persist for more than one day. If the infant is otherwise well, no investigation or treatment is required, but recommendation for review should be made if the rash does not resolve after one to two weeks. Milia, which are a result of blocked pores, typically present as tiny, white cysts on the face in about 40–50% of newborns. Most lesions resolve after one to two months. Parents should be reassured and advised not to squeeze or pick them as this may result in infection or scarring.

Conclusion

Neonatal assessment can be challenging because of the non-specific nature of presenting signs and symptoms. A thorough clinical assessment, including relevant maternal medical, antenatal, peripartum and postpartum histories, with a systematic physical examination, is the most important tool in the primary clinic setting.

References


