



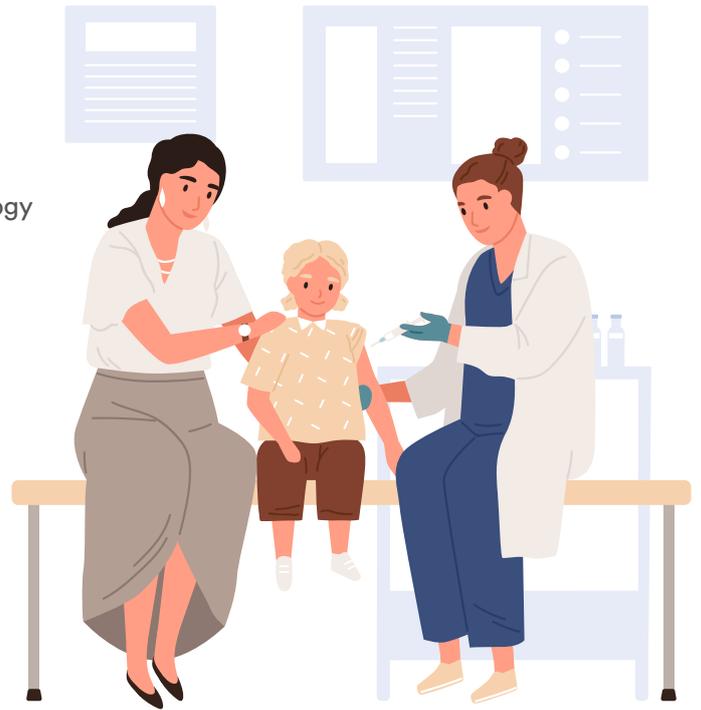
**Student Section  
Paediatrics Specialty  
Educational Booklet**

**November 2020**



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## Notes from the Editors

During this unprecedented time, hospitals and NHS staff have had to significantly change their way of working; whether that is through remote consultations, increased PPE or adapting to strained resources. This has been especially challenging in the field of paediatrics, where parents have become reluctant to bring their children to hospitals and emergency departments in fears of COVID-19.

The pandemic has also drastically changed medical student learning; placements are now restricted for certain clinical years to prevent their exposure to potentially 'red' zones and to comply with social distancing regulations. Exams have either been rescheduled or cancelled for some students across the country. In this booklet, we have included some revision resources for medical students to use on their paediatrics placements (pages 10, 12, 14). For students who have had their electives cancelled, you can immerse yourself in the experience of Dr Alisha Burman who undertook a thoroughly valuable paediatrics elective (page 8). To delve deeper into the basic science of some important conditions in paediatrics, we have two articles exploring important genetic disorders (pages 6, 12).

It is important to remember that medicine encompasses far more than diseases but is also holistic which is covered in our article regarding mental health in children and adolescents (page 3). It is also extremely important to consider ethnic representation in dermatology, and this is covered in page 4.

I hope that you will be inspired about a career and paediatrics and learn a lot from this booklet to inspire confidence going into your exams and OSCEs.

On behalf of the RSM Student Section Core Committee,

Maiar Elghobashy, University of Birmingham

Editor

## Why paediatrics?

Children have always taken a special place in my heart. I find them to be great teachers of life, with endless learning experiences whilst you are around them. It was on a humanitarian trip to Kenya where I worked with children that my love for them was truly captured. After having spent an entire day at an orphanage in Kenya, I found myself with an overwhelming urge to redirect my purpose into caring for the lives of children. It was at this point that I knew I wanted to pursue a career in paediatrics.

Paediatrics appeals to me because it is a speciality where I can continuously advocate for children's health and safeguard them. It is a speciality where I feel I can make the most difference. There is also something so special and rewarding about positively impacting a child's life, as well as their parents' lives.

With the experiences, I have gained in recent years, I have witnessed how a paediatrician is present in one's life from birth to adulthood. Taking that journey together with a child is another aspect of paediatrics that interests me. A child tends to be full of wonder and happiness, whilst making their day better, they are also making a difference in your day. Children also often portray the adults around them as heroes and I would one day like to be a doctor heroine to them.

Victoria Khaukha, University of Buckingham

Editor

# Child and Adolescent Mental Health (CAMHS)

Victoria Khaukha, University of Buckingham  
Dr Cate Manning, Great Ormond Street Hospital

## Why should you choose CAMHS?

There are few specialities in paediatrics that offer as much variety as CAMHS. You can go from seeing a teenager in A+E who has self harmed after being bullied, to assessing a young boy who keeps getting into trouble at school and cannot seem to connect with his peers, to carrying out a Mental Health Act assessment on an adolescent who has been brought to hospital by the police with a relapse of her psychotic illness. In community CAMHS, presentations range from depression and anxiety, to eating disorders, autism, behavioural difficulties and severe and enduring mental illnesses. Whilst most CAMHS consultants will specialise in a particular area, even within these subspecialties there is huge variety in presentations. For instance, in Paediatric Liaison, you can find yourself on any ward in the hospital, dealing with depression in the context of a chronic illness, acute behavioural disturbance secondary to delirium, or psychosomatic disorders where the physical symptoms may have a strong psychological component.

One of the great joys of working in CAMHS is that we approach the patient as a person, and we need to understand their whole lives and not just the symptoms that have brought them to us. It is a huge privilege to hear their stories and it is impossible to be bored. Arriving at the correct diagnosis – and there is often more than one – is a skilled and sometimes lengthy task. You have to have the patience to deal with uncertainty for long periods of time and the flexibility to keep an open mind even once you've made a diagnosis, in case new symptoms emerge. Accurate diagnosis is important and satisfying – as it will guide the evidence-based treatment. One of the most rewarding aspects of child mental health work is that we have some of the most effective treatments in the whole of medicine. For example, 80% of children treated for anxiety have good responses, and effective management of attention deficit hyperactivity disorder can transform a child's educational and social trajectory. You have to employ a range of problem-solving skills that often require an imaginative and thoughtful approach. From helping a young person to identify distraction techniques for when they feel like self harming, to creating an environment and context that helps a young person feel safe and able to confide deeply personal matters or tackling ethical questions such as whether it is in a young person's best interests to be detained under the Mental Health Act, there is an opportunity to use all the critical thinking skills you learn at medical school. When this approach is paired with compassion and a capacity to take an overview of the whole system around the child, then you're thinking like a Child Psychiatrist.

## Why is CAMHS important?

Mental illnesses affect an individual's day to day life tremendously. The most recent national survey of children's mental health (The Mental Health and Children and Young People, MHCYP 2017<sup>1</sup>), showed that 12.8% of 5 to 19-year olds have a mental disorder. As a comparison, around 10% of children between 0-15 years old are affected by asthma<sup>2</sup>.

The high rate of mental disorders in children is a serious health



and social problem. As well as measuring the prevalence of mental disorders, the MHCYP survey also highlighted their consequences across multiple domains of a young person's life:

- Truancy levels were ten times higher in those with mental disorder (8.5% vs 0.8%)
- Exclusion was 13 times higher in those with mental disorders (6.5% vs 0.5%)
- Illicit drug use was found to be three times more likely in 11 to 16-year olds with a mental health condition (13.9%) than those without
- 11-19-year olds with a mental disorder were nearly twice as likely to have been bullied in the past year as those without (59.1% vs 32.7%), and nearly twice as likely to have been bullies themselves
- 71.7% of children with a mental disorder had a physical health or developmental condition and 25.9% had a limiting long-term illness compared with 4.2% of children without a mental disorder

With the wide-reaching effects of mental illness laid bare, it becomes clear that treating mental illnesses can impact hugely on a child's life. There are few things more satisfying than helping to change the trajectory of a child's life towards accessing better educational opportunities, forming positive social relationships and feeling hopeful about their future.

There is increasing recognition by the government that there is a huge unmet need for mental health interventions within our young population. The world in which children are now growing up is increasingly challenging thanks to social media, climate change, political instability and the impact of the Covid-19 pandemic. Working in CAMHS offers an opportunity for imaginative, passionate and dynamic doctors to help our specialty to re-imagine the way we operate and to bring together education, social care and communities to work with us to meet the growing social, emotional and psychological needs of young people.

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# Addressing Ethnic Representation in Paediatric Dermatology

Caterina Shepherd, University of Buckingham

Over the past few months there has been increasing awareness over the lack of representation of ethnically diverse skin types in dermatology resources. This comes at a time when the COVID pandemic and the Black Lives Matter movement has highlighted the urgent need to address health inequalities among ethnic minorities. In June 2020, a petition was launched calling on the British Association of Dermatologists (BAD) to update teaching resources to include examples and diagnostic criteria relevant to patients of Black, Asian and Minority Ethnicities (BAME) [1]. Following widespread support, the BAD responded that they are in the process of improving resources available [1]. However, before the changes come into effect, it is important for clinicians and medical students to be aware of limitations in current resources and take measures to mitigate the lack of ethnic representation on diagnosis and the treatment of ethnic minorities.

## Health inequality

The lack of ethnic representation in medical literature has been identified as a key contributor towards the disproportionate burden of disease in ethnic minorities. Clinicians and medical students have reported feeling unconfident in identifying signs of skin conditions that present differently in BAME patients compared to Caucasian patients [2]. These include jaundice, pallor, blanching and cyanosis, which can all be influenced by skin colour [3]. Of particular concern are potentially life-threatening conditions such as melanoma. Ethnic minorities are thought to be three times more likely to die from melanoma as individuals from non-minority groups [2]. Lack of BAME relevant training in the medical profession is considered a key contributor to this disparity as it contributes to reduced detection rates and misdiagnosis [4]. Skin colour also affects a range of other skin characteristics with dermatological implications (see table below [4]).

## Characteristics of ethnic skin and their effects. Adapted from Vashi et al. [4]

CHARACTERISTIC	DERMATOLOGIC IMPLICATION
Increased tyrosinase activity leading to increased melanin content	Greater photoprotection leading to decreased incidence of skin cancer
Larger melanosomes	Less noticeable photoageing
Labile melanocytes and slower melanin degradation	Dyschromias (alteration of colour of skin)
Thick dermis	Preserved skin elasticity for a longer period of time
Larger multinucleated fibroblasts	Greater prevalence of hypertrophic scarring and keloids in coloured skin

## Skin colour considerations in serious paediatric dermatological conditions

### Meningitis

Meningitis is the leading infectious cause of death in children in the UK [5]. A key diagnostic feature on fair skin is the presence of a non-blanching, petechial rash. This presentation, described as ‘classic’ by the BMA, is harder to observe on darker skin [5]. Clinicians should inspect paler areas, especially the conjunctivae, soles of the feet and palms of the hand to ensure they do not miss erythema in darker skinned children [5]. Furthermore, pallor is considered a classic sign of clinical deterioration in meningitis. This symptom is less easily detected in darker skinned children. Clinicians should ensure they assess for pallor in areas where skin tone is lightest including the conjunctivae, mucous membranes and nail beds [3].

### Melanoma

Melanoma is considered the deadliest form of skin cancer in children and adolescents [6]. It arises from uncontrolled proliferation of melanocytes [7]. Although non-Caucasian ethnic groups have a reduced risk of developing melanoma, they are more likely to be diagnosed at a later stage and have higher mortality rates [8, 9]. The main reasons for this phenomenon described in the literature are summarised below.

In BAME individuals, melanoma often develops in sites that have low sun exposure such as the soles of the feet and palms of the hands (acral lentiginous melanoma). These sites are more likely to be missed on visual examination resulting in delayed detection and poorer prognosis [8].

Colours for melanoma described in the literature typically include tan, dark brown or black. It can be harder to identify melanomas or precursor lesions in dark skin as the surrounding skin may match or camouflage the colour of the melanoma [2, 8, 9].

The ABCD classification of skin cancer generally features white skin and not skin of darker ethnicities [10]. This may reinforce the concept that skin cancer is less prevalent in darker skinned patients and may contribute to a low index of suspicion in both clinicians and patients.

### Conclusion:

There is growing awareness that leading dermatology resources available to clinicians and patients do not adequately represent ethnic differences in disease presentation. Ethnic differences are often overlooked or referred to as “atypical” which can not only affect health outcomes through reduced detection and diagnosis, but also affect the psychosocial wellbeing of minority ethnic patients by contributing towards feelings of alienation and exclusion. Timely diagnosis and ethnic and culturally sensitive support resources could mitigate this and thus further research is urgently needed to explore this topic further.

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# Prader Willi and Angelman Syndrome: Similar but different

Mirna Elghobashy, University of Birmingham

Angelman Syndrome (AS) and Prader-Willi Syndrome (PWS) are two neurodevelopmental genetic disorders arising from the same area on the long arm of chromosome 15 (1). Despite this, their clinical presentations are very different. PWS primarily results in hypotonia and hyperphagia, while AS causes cognitive impairment. This region on chromosome 15, specifically 15q11-q13, is of particular interest as it is subject to genomic imprinting; a process by which genes from parent alleles are tagged and only one set is expressed preferentially. This is through various epigenetic mechanisms, such as DNA methylation and histone modification, which determine whether a gene is transcribed and, therefore, expressed (2). Defects of this process can result in inappropriate silencing of a gene, although normal chromosomes are present; these are known as imprinting mutations.

AS primarily affects the nervous system, causing developmental and physical abnormalities (4). It is estimated that AS affects 1 in 20,000 people worldwide, classifying it as a rare disease. AS occurs as a result of loss of function of the UBE3A gene (4). Only the maternal copy of this gene is expressed in the brain while the paternal gene is always deactivated epigenetically. Therefore, a mutation or deletion of the maternal gene, paternal uniparental disomy (two copies of the gene from the father) or imprinting defects (inappropriate silencing of the maternal gene) will result in the characteristic symptoms of AS (4). Typical symptoms of AS include cognitive impairments, ataxia and absent speech. Patients may also exhibit microcephaly and delayed development. Epilepsy develops in 85% of AS patients and the most common types of seizures are atypical absence or generalised tonic-clonic (5). These symptoms usually become evident by twelve months of age although some manifestations, such as seizures, may present later. Children with AS typically have a happy and excitable attitude although this tends to decrease with age (4). There is no standard treatment for AS, and current practice involves managing and alleviating major symptoms as well as behavioural and communication therapy (4). This includes anti-epileptic medication, physical therapy and speech therapy with an emphasis on communication aids and signing (6).

Prader-Willi syndrome is another genetic disorder arising from various genes in the 15q11-15q13 region. It can occur as a result of deletions of the paternal copy of this region, maternal uniparental disomy or imprinting defects resulting in loss of paternal gene expression (7). PWS results in infantile hypotonia, failure to thrive and cognitive impairment. Many manifestations of PWS include hypothalamic and gonadal insufficiency (7). This can result in underdeveloped sex organs, incomplete development at puberty and infertility. Due to a growth hormone insufficiency, children with PWS often have short stature and relatively decreased muscle mass(8). As affected children often do not feel satiated following eating, without intervention they can develop life-threatening obesity due to overeating (8). Typical facial features include narrow foreheads, almond shaped eyes and triangular mouths. There is no cure for PWS, and management depends on the severity and variation of symptoms. In infancy, nasogastric tubes or special nipples may be used if hypotonia compromises the baby's feeding

ability (8). It may also be beneficial to use hormone therapies to compensate for hypothalamic insufficiency, such as growth hormone. Behavioural therapy may prove useful for managing patients' diets and childhood supervision of food intake is necessary to prevent excessive weight gain(8).

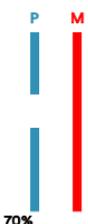
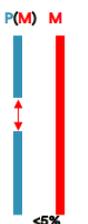
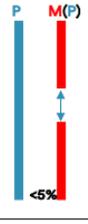
	Deletion	Uniparental disomy	Gene mutation	Imprinting mutations
Prader-Willi Syndrome	 70%	 25%	 0%	 <5%
Angelman Syndrome	 70%	 2%	 20%	 <5%

Table 1. Causes of PWS and AS and their relative percentages. Note that uniparental disomy accounts for more cases of PWS than AS due to the high rate of maternal nondisjunction. Blue represents the paternal allele of the long arm of chromosome 15 while red represents the maternal. (9)

Gene imprinting is an important mechanism in humans on a genomic and phenotypic level, allowing accurate prediction of the likely consequences of deletions and mutations in DNA sequences. While AS and PWS arise from the same region of chromosome 15, the effects of gene imprinting mean that their signs and symptoms are very different. It is, therefore, important to be aware of both disorders and their clinical presentations. The mechanisms of imprinting are not fully understood, and more research and experimentation will inevitably lead to positive outcomes for patients. Knowledge of genomic imprinting can influence treatment and provide novel theories and pharmacological interventions for symptomatic relief of genetic disorders.

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# Antenatal repair: The future of spina bifida treatment?

Daniel Richardson, St George's University

## Overview:

Neural tube defects encompass a broad range of birth defects that can affect the brain or spinal cord and occur as a result of abnormal neurulation or closure of the neural tube at around week 4 of embryological development<sup>1</sup>. These abnormalities range from the clinically insignificant spina bifida occulta to conditions that are incompatible with life, such as anencephaly. Therefore, the majority of treatments are focused on conditions in the center of this spectrum where maximal improvement to quality of life is possible.

The two conditions where treatment is therefore predominantly focused upon are meningocele's and myelomeningocele's, where the meninges, or in the latter case both the meninges and the spinal cord are exposed. These conditions are often collectively referred to as 'open spina bifida'<sup>1</sup>.

## Treatment:

In the majority of cases, spina bifida is detected antenatally and historically repair of 'open' spina bifida has occurred in the days following birth to prevent meningitis and provide a cosmetic repair of the defect<sup>1</sup>. Those born with 'open' spina bifida often experience variable paralysis and sensory loss in the legs as well as bowel and bladder problems depending on the level of the lesion.

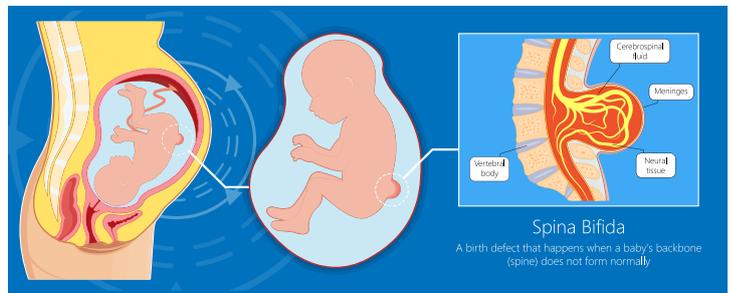
The Management of Myelomeningocele Study (MOMS)<sup>2</sup>, a randomised clinical trial, was undertaken to investigate whether prenatal intervention conferred any benefit over postnatal closure. The study was stopped on efficacy grounds, with significantly reduced rates of death and need for CSF shunting at 12 months in patients treated antenatally. A further outcome measured (mental development and motor function) was also improved at twelve months.

Unfortunately, whilst foetal outcomes were improved, antenatal interventions subject mothers to risky surgery that they would otherwise not experience<sup>3</sup> and as a result, the rate of complications were higher for both mother and baby. Such complications included increased rates of preterm labour and rupture of the uterus in subsequent pregnancies. Furthermore, the MOM study was conducted at highly specialised institutions by experienced surgeons and used an extremely stringent selection criteria, which does not reflect standard care<sup>3</sup>.

Whilst the MOMS trial demonstrated the potential of antenatal intervention, it did not provide such a clear-cut benefit as had been hoped<sup>3</sup>. As a result, the majority receive prophylactic intravenous antibiotics and post-natal repair during in the first few days of life.

## Recent Developments:

In 2019 there were 23 centres offering antenatal repair of spina bifida worldwide, 6 of these based in Europe<sup>4</sup>, demonstrating a slow but steady increase in centres offering antenatal interventions.



To address the poor maternal outcomes associated with open repair of spina bifida, fetoscopic (keyhole) approaches to antenatal repair of spina bifida have been attempted. Whilst these show promise and theoretically could decrease the rate of maternal complications, no standardised approach and technique yet exists<sup>4</sup>.

Last year the National Institute of Clinical Excellence approved funding of 'open' and 'fetoscopic' antenatal spina bifida repair in specialist institutions<sup>5</sup>.

## Future research considerations:

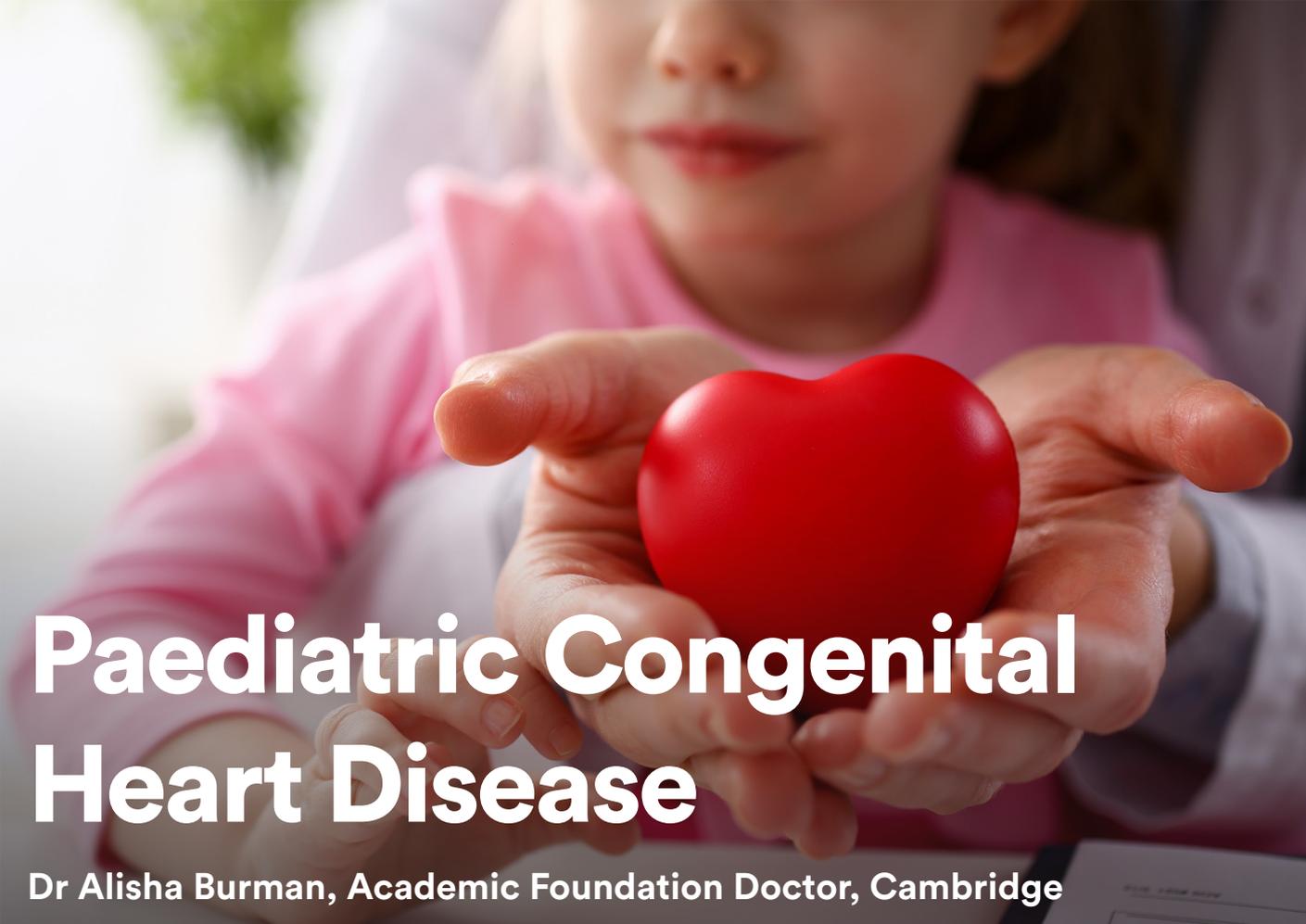
There is no doubt that antenatal intervention of spina bifida will continue to be adopted and refined, especially in respect to fetoscopic approaches<sup>4</sup>. Surgical robots, offering 3D optics and advanced articulated instruments, have previously demonstrated feasibility through animal trials<sup>6</sup> though human trials of these technologies have yet to be undertaken and little further research on robotic-assisted fetoscopy has been published in recent years.

Robotic surgery has, however, been used in pregnancy previously for both obstetric indications, including excision of pelvic masses, endometrial tumours and cerclage<sup>7, 8</sup>, and non-obstetric reason including adrenalectomies<sup>9</sup>. The successful outcomes associated with these procedures demonstrate that robotic surgery can be performed safely during pregnancy.

Antenatal surgery for spina bifida is currently the only non-lethal condition for which surgery of this nature is currently offered. As outcomes continue to improve, we may see more procedures that were historically were performed during the neonatal period, performed antenatally.

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# Paediatric Congenital Heart Disease

Dr Alisha Burman, Academic Foundation Doctor, Cambridge

## Introduction

Paediatric cardiology is a subspecialty that you may not get much experience of while being a medical student. During my rotation in paediatrics, I was fortunate to witness antenatal counselling for an unborn child with congenital heart disease. The clinicians discussed how they planned to manage the congenital heart disease once the child was born. This piqued my interest and I was keen to learn more about paediatric cardiology and cardiothoracic surgery. As a result, I organised placements in Great Ormond Street Hospital and Vietnam.

## My experiences

In Great Ormond Street Hospital (GOSH), I spent time with the cardiothoracic surgeons and anaesthetists, witnessing specialised surgery such as the Fontan procedure. The intricacy and the variety of operations amazed me, and I saw them operate on tiny babies to older children. I felt incredibly lucky to witness the operations and the team were kind enough to provide teaching and explain what was going on during the operation.

## Vietnam

During my medical elective, I worked at a large tertiary hospital in Vietnam in the paediatric cardiothoracic department. Here, I was able to spend time with cardiologists, surgeons and intensivists, who worked together to manage the child's congenital heart disease. This was incredibly valuable as I got to witness their care from different perspectives. During the placement, I was able to

clerk pre-operative patients, observe surgeries procedures in the catheter lab and assist in managing post-operative patients in intensive care. I was also taught how to do transthoracic and transoesophageal echocardiograms and how to interpret them.

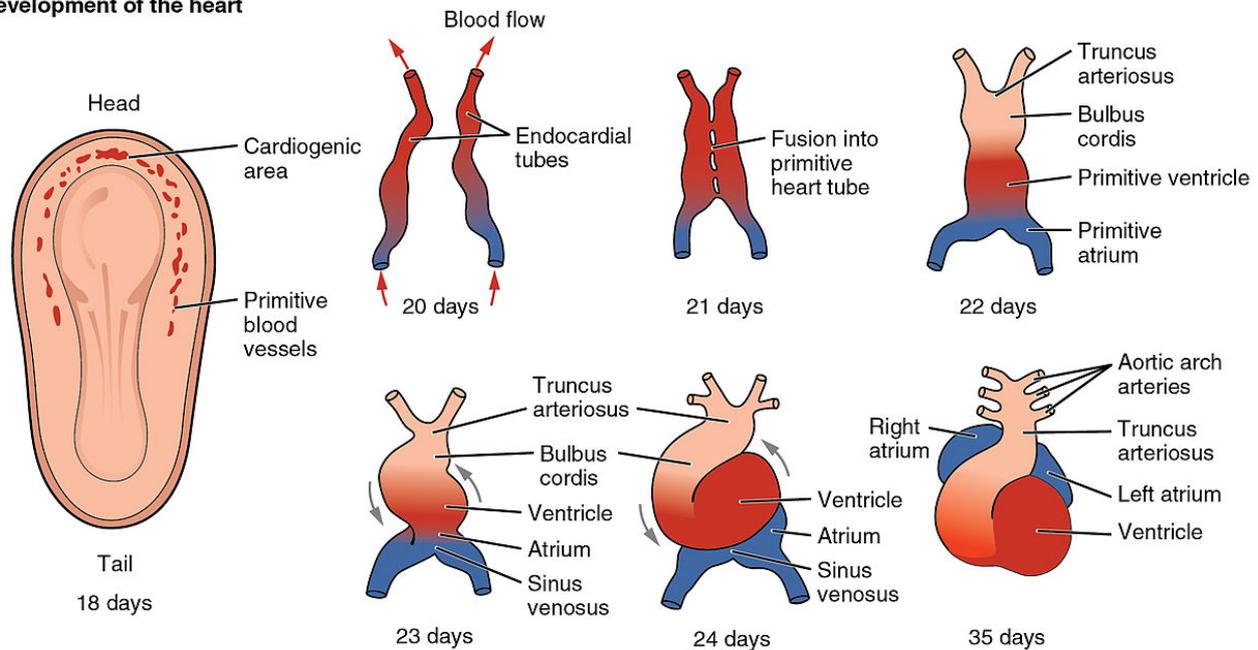
It was interesting to compare and contrast my experiences in Vietnam to that in Great Ormond Street hospital. One of the most striking things was that the children, who did present to hospital, presented later in Vietnam. They found that quite a few children were being identified too late and already had pulmonary hypertension, making surgical management not a viable option. As a result, the Vietnamese doctors have to be inventive with the resources available. The paediatricians would travel to rural areas every other month with the aim of identifying children earlier and planning how to manage the child's congenital heart disease with their families. They are also training doctors in rural hospitals in how to manage these children with the resources available to them. This will allow others to set up paediatric cardiothoracic centres and families will not have to travel so far to access the care they need.

## Embryology of the heart made simple

There are many different pathologies that come under the umbrella of congenital heart disease. In order to understand the pathophysiology underlying congenital heart disease, it is important to have a grasp on normal heart development.

The embryo has 3 germ cell layers: ectoderm, mesoderm,

## Development of the heart



Primitive Heart Tube	Developed Heart
Truncus Arteriosus	Aorta and Pulmonary artery
Bulbus Cordis	Right Ventricle
Primitive Ventricle	Left Ventricle
Primitive atrium	Anterior part of left and right atria and their appendages
Sinus Venosus	Posterior part of right atrium, sinoatrial node and coronary sinus

and endoderm. The heart arises from the mesoderm which differentiates further into mesothelium, endothelium, and myocardium. The mesothelium will become the pericardium and the endothelium will become the endocardium, which forms the inner lining of the heart.

By day 19 of foetal development, there are two endocardial tubes, which merge to form a singular tubular heart by day 21. This is known as the primitive heart tube. At day 22, it differentiates further into distinct areas: the truncus arteriosus, bulbus cordis, primitive ventricle, primitive atrium and sinus venosus. Contraction of the primitive heart tube begins at day 22 and allows blood to flow from the sinus venosus to the truncus arteriosus. The parts of the heart tube will eventually form the key parts of the developed heart (see Table 1). The truncus arteriosus divides into the aorta and pulmonary artery. The bulbus cordis becomes the right ventricle and the primitive ventricle develops into the left ventricle. The primitive atrium forms the atrial appendages and anterior part of the right and left atria and the sinus venosus will become the posterior part of the right atrium, sinoatrial node and coronary sinus.

At day 23, the primitive heart tube continues to elongate and begins to fold to morph into a shape resembling a fully developed heart. The bulbus cordis moves anteriorly, caudally and to the right and the primitive ventricle moves

posteriorly, cranially, and left. This looping of the heart tube finishes by day 29 and there is now left to right asymmetry in the thorax.

The sinus venosus receives venous blood from both the right and left sinus and empties the blood into the primitive atrium. During the fourth week, venous return shifts to the right side of the heart and the right sinus is engulfed into the right atrium, forming the inferior vena cava. The left sinus forms the coronary sinus, which drains venous blood from the myocardium into the right atrium.

Septation begins during the fourth week. Endocardial cushions that develop in the atrioventricular region grow and will aid septation of the heart. In the atrium, the septum primum (first septum) extends to the endocardial cushions to split the atrium into the right and left atria. Before fusion is complete, the hole present in between the septum primum and endocardial cushions is known as the ostium primum (first opening). The ostium primum is eventually closed with the growth along the septum primum. When this happens, a second opening known as the ostium secundum is formed within the septum primum. A second septum grows (septum secundum) which has a hole known as the foramen ovale. The septum and their openings allow for right to left shunting of blood throughout the development of the heart, so the blood can bypass the foetal lungs.

The ventricles are divided by the interventricular septum, which contains a muscular and membranous portion. The muscular part grows upwards from the base of the ventricles towards the endocardial cushions; however, it leaves a small opening known as the primary interventricular foramen, which is covered by the membranous portion of the interventricular septum.

## Conclusion

My experiences in paediatric cardiology have been varied and have encouraged me to delve beyond the medical school curriculum to explore an area that I am interested in. Regardless of specialty, I think if you want to get more experience as a student, most clinicians would be happy to be contacted and may even advise you on how to gain more experience.



# Paediatric Oncology: 3 childhood cancers to consider in your differentials

Chris Smith, University of Southampton

It's news that no parent wants to hear, but a differential that no medical student can afford to forget. Cancer is diagnosed in around 5 children a day in the UK<sup>1</sup>, with leukaemias, lymphomas and CNS malignancies making up over two-thirds of cases<sup>1</sup>. Children's cancers often present with common, non-specific symptoms so it's important to recognise which red flags need to be investigated further.

## Leukaemia

Acute Leukaemias are the most common childhood cancer, particularly Acute Lymphoblastic Leukaemia (ALL)<sup>2</sup>. Signs and symptoms are either a result of bone marrow dysfunction caused by the leukaemia or the deposition of leukaemic collections around other organs. In the case of the former, a lack of erythrocytes can lead to fatigue, weakness, pallor or shortness of breath<sup>2</sup>, whereas a lack of platelets can lead to a child bruising easily or having bleeding gums and severe or frequent nosebleeds<sup>2</sup>. Though a child with leukaemia will have a high overall white cell count, these will be predominantly immature malignant cells so frequent infections may occur as a result of neutropenia<sup>2</sup>.

The most common clinical findings in children with

leukaemia are hepato- and splenomegaly<sup>3</sup>, either separately or combined. This is caused by a build up of leukaemia cells around the viscera. If a large mass develops at the thymus, it can obstruct the vena cava and lead to the life-threatening "SVC-Syndrome" which causes headaches, loss of consciousness and bluish-purple swelling of the upper body.

Apart from leukaemia, the symptoms of pallor, shortness of breath and fatigue can be caused by a variety of other anaemias, for example iron deficiency or haemolytic anaemias. Increased bruising could be due to non-accidental injury and recurrent infections could be due to an immune deficiency.

To investigate a possible diagnosis of leukaemia, a full blood count should be taken to assess the number of erythrocytes, platelets and white cells, and a blood film should be performed<sup>4</sup>. On a blood film, leukaemia is characterised by a high number of poorly-differentiated blast cells or leucoerythroblastosis<sup>4</sup> - a high number of immature, nucleated erythrocytes. Bone marrow biopsies are used to confirm the diagnosis after laboratory analysis. In stark contrast to solid tumours, imaging is not particularly useful when investigating leukaemia as discrete masses do not form<sup>4</sup>, but chest x-rays can be used to detect an enlarged thymus or hilar lymphadenopathy<sup>2</sup>.

## Lymphoma

Depending on the age of the child or young person, different types of lymphoma are more common; Hodgkin's Lymphoma is the most common cancer in 15-24-year olds<sup>5</sup>, but non-Hodgkin lymphoma is more common in children younger than 14<sup>6</sup>. Both types are extremely uncommon in children under 3<sup>6</sup>. Symptoms of lymphoma in children are non-specific; a painless, persistent lymphadenopathy is a common sign on presentation<sup>7</sup>, as are extreme fatigue and generalised itching<sup>7</sup>. Collectively the triad of drenching night sweats, unexplained weight loss and fevers are known as "B symptoms" and are highly suggestive of a lymphoma<sup>7</sup>. In the case of Burkitt's lymphoma, children can present with abdominal distention and vomiting, or toothache and jaw pain if the mandible is affected<sup>7</sup>.

If lymphoma can be ruled out, other differentials for the clinical picture described above can include cytomegalovirus, infectious mononucleosis or tuberculosis. Itching could be caused by a dermatological condition such as eczema, and abdominal distension could be caused by a condition such as coeliac disease.

Diagnosis of a lymphoma can be confirmed by analysing a lymph node biopsy, the results of which may be supplemented by a chest X-ray or CT to determine the position and size of any enlarged lymph nodes, which in turn determines the staging of the disease<sup>8</sup>. Full blood counts can be used to assess whether the lymphoma has infiltrated the bone marrow and is suggested by a pancytopenia<sup>8</sup>. Lymphoma in children generally has an excellent prognosis, with around 80-95% of children surviving after 5 years<sup>6</sup>.

## Brain Tumours

The final category of childhood cancers is brain tumours. They are the most common solid tumours in children, and prognosis varies dramatically based on what cell the malignancy arises from. Similarly, the signs and symptoms

on presentation vary depending on the tumour's location but many present with common symptoms such as nausea, vomiting or a recurring headache<sup>9,10</sup>, which are all signs or raised intracranial pressure<sup>11</sup>.

Medulloblastomas, which account for 20% of paediatric brain tumours<sup>10</sup>, grow in the posterior fossa and can cause inattention, clumsiness and incontinence of bladder and bowel if there are spinal metastases<sup>10</sup>. Other groups of tumours form supratentorially and can lead to focal neurological deficits, decrease in cognitive ability or delay in reaching milestones<sup>9</sup>. In younger children this can present as irritability or drowsiness. A final example of the diverse range of paediatric brain tumour signs is visual disturbance. This occurs when a glioma infiltrates the visual pathway and results in a presentation of nystagmus, proptosis, strabismus or loss of vision<sup>10</sup>. Visual disturbance is the most common presentation in children with brain tumours under 4<sup>11</sup>.

Differential diagnoses to consider alongside brain tumours include primary headache, epilepsy or a behavioural disorder such as ADHD, depending on the presenting signs and symptoms.

To confirm that a brain tumour is present, imaging is essential- MRI and CT of the head can be used to determine the tumour's location and the spine can also be imaged if metastasis is suspected. Fundoscopy to detect papilloedema is essential for early diagnosis and lumbar punctures can determine whether tumour cells are circulating in the cerebrospinal fluid. Definitive diagnosis of tumour type is by biopsy during neurosurgical tumour resection.

### But remember: Paediatric Cancer is Rare!

Though they're important to remember, the adage "common things are common" is particularly relevant when ruling out childhood cancers; it's estimated that the average GP will only encounter a new paediatric malignancy once every 20 years<sup>12</sup>. Despite this, by ensuring that these diagnoses are recognised and considered you're not only showing any examiner that you're aware of paediatric oncology presentations, you might also change a child's life.

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# Common paediatric genetic conditions

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## DOWN'S SYNDROME

### *Summary of condition*

Down's syndrome is caused by trisomy 21. This alters the development and growth of the baby and poses both mental and physical challenges. It is associated with intellectual disability and characteristic facial features.<sup>[1]</sup> Common features include:

- Flat nasal bridge, epicanthal folds
- Low muscle tone
- Shorter height <sup>[2]</sup>

Typical presentation of Down's syndrome in a neonate <sup>[2]</sup>

### *Diagnostic methods:*

Early detection is possible via screening and diagnostic tests. Screening tests consists of an ultrasound scan and a blood test. Diagnostic tests including chorionic villus sampling and amniocentesis and percutaneous umbilical blood sampling. <sup>[2]</sup> <sup>[3]</sup>

### *Treatments available:*

Management strategies for individuals with Down's syndrome include speech (improves communication), occupational (builds motor tone and eases daily tasks) and behaviour therapies (helps manage the emotional struggles). <sup>[3]</sup>

**UK Statistics** - 1 in 1000 babies born in the UK. <sup>[4]</sup>

## FRAGILE X SYNDROME

### *Summary of condition*

Fragile X Syndrome is an X-linked disorder caused by a mutation in the FMR1 gene. <sup>[5]</sup> Individuals affected experience various characteristics, some of which include:

- Autistic-like features i.e. avoiding eye contact and anxiety in social situations
- Mild to moderate learning disabilities
- Speech and language delay <sup>[5]</sup>

They also exhibit some key physical features including:

- Macrocephaly
- Prominent ears and jaw
- Macroorchidism <sup>[5]</sup>

### *Diagnostic methods:*

Diagnosis is confirmed by molecular genetic testing of blood DNA. Results are usually available in 6 to 8 weeks in the UK. <sup>[6]</sup>

### *Treatments available:*

Treatments available are focused on improving coping mechanisms and facilitate day-to-day activities. They include special education, sensory integration training as well as therapy. In terms of medication, SSRIs, antipsychotics and anticonvulsants are prescribed for symptoms of anxiety, mood stabilisation and seizures respectively. <sup>[5]</sup> <sup>[7]</sup>

**UK Statistics** - 1 in 4000 males and 1 in 8000 females in the UK. <sup>[5]</sup>

## CYSTIC FIBROSIS

### *Summary of condition*

Cystic Fibrosis is an inherited disease caused by a mutation of the CFTR gene inhibiting the movement of chloride to the cell surface. Mucus becomes thick and sticky and clogs the lungs thus increasing the risk of infections, respiratory failure along with a plethora of complications. <sup>[8]</sup>

### *Symptoms:*

- Persistent cough
- Male infertility
- Poor growth <sup>[8]</sup>

### *Diagnostic methods:*

Diagnosis includes genetic testing i.e. blood or saliva for mutation of CFTR gene, sweat test, and a clinical evaluation at the Cystic Fibrosis Foundation-accredited care centre. [9]

**Treatments available:**

Treatment includes bronchodilators to relieve shortness of breath, antibiotics to control persistent infections, mucolytics to break down the mucus as well as physiotherapy to clear the lungs of the mucus. [8][9]

**UK Statistics** - 1 in every 2,500 babies born in the UK. [9]

## SICKLE CELL ANAEMIA

**Summary of condition**

It is a disorder caused by a mutation in the HBB gene which codes for haemoglobin. This changes the normal round shape of haemoglobin into a new form called HbS. HbS has a characteristically sickle shape, making it more likely to block vessels and damage blood vessels. [10]

Serious clinical consequences include:

- Infections e.g. meningitis
- Pulmonary hypertension
- Delayed puberty [11]

An illustration of healthy and sickled cells [10].

**Diagnostic methods:**

Diagnosis is confirmed using haemoglobin electrophoresis. It is offered to the mother prior to 10 weeks gestation. [10]

**Treatments available:**

Regular blood transfusions can also help reduce the risk of complications. Regular antibiotic medication also protects against chronic infections in children below the age of 5. [10] [11]

**UK Statistics** - 1 in 76 babies born in the UK will carry sickle cell trait. [10]

## TURNER SYNDROME

**Summary of condition**

Turner Syndrome is a genetic disorder caused by loss of part of all of the X chromosome and only affects females. [12] The disorder varies from individual to individual, but individuals have distinct features, such as:

- Failure to grow i.e. short stature
- Webbed appearance of neck
- Drooping eyelids [13]

**Diagnostic methods:**

A diagnosis is confirmed by chromosomal analysis using karyotyping. [12] [13]

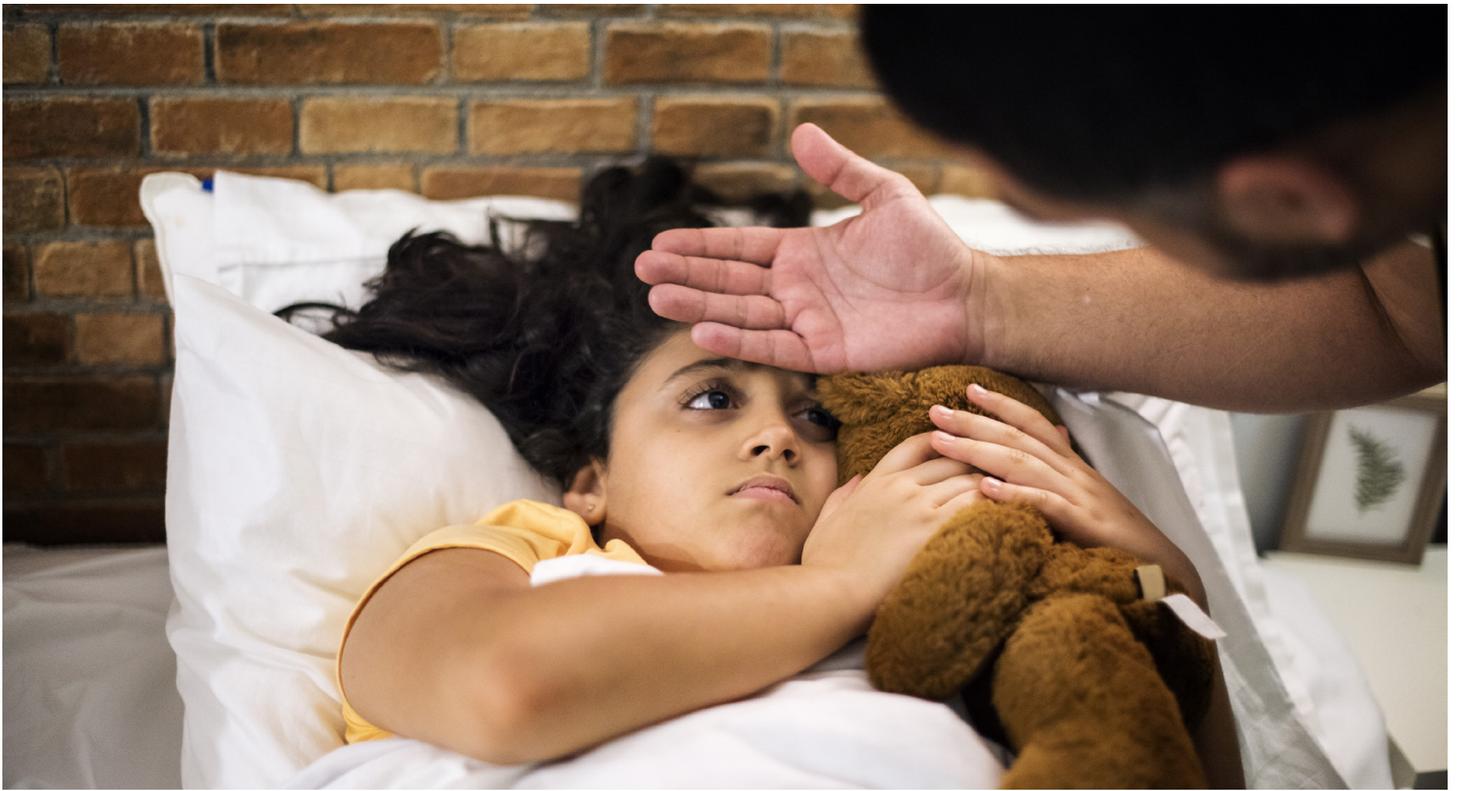
**Treatments available:**

Individuals are often required to attend regular health checks with a number of healthcare professionals including paediatric endocrinologists and psychologists. Oestrogen and progesterone replacement therapy is often recommended as it promotes sexual development and prevention of osteoporosis. [12] [13]

**UK Statistics** - 1 in every 2,000 baby girls in the UK. [13]

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# Spotting the sick child

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## Why is this important?

Children often present with vague symptoms and find it difficult to communicate how they are feeling thus early identification of spotting a sick child is extremely important.

## THE 3 MINUTE TOOL KIT (ABCD ENT TT)

### INTRO

- Wash hands
- Introduce yourself
- Patients name and date of birth
- Explain examination
- Reposition patient

## Airways → can become easily blocked

Listen for:

- Secretions
- Stridor
- Foreign body

## Breathing

- Respiratory rate → to assess if in respiratory distress or systemic problems e.g. DKA
- Accessory muscle use
- Oxygen saturation → important early warning sign
- Auscultation → e.g. listen for wheezing, crepitations, bronchial breathing

## Circulation

- Colour → if pale, mottled arms or legs (*could indicate poor perfusion*)
- Heart rate
- Capillary refill time
  - Measure centrally: by pressing on sternum
  - Peripherally: by pressing on finger, toes, hands or feet
- Temperature of hands and feet (*compared to trunk*)

## Disability

- Pupils e.g.
  - Sluggish reflex: after fit or drug overdose
  - Changing pupil sizes: could indicate ongoing seizures
  - Asymmetry pupils: space occupying lesion in brain e.g. extra or subdural haemorrhage post head injury
- Limb tone & movement
  - important to compare if you are worried about a space occupying lesion in brain e.g. haematoma or tumour
- AVPU (*alert, verbal, pain, unresponsive*) or GCS (*Glasgow coma scale*) score

## ENT

- Examine if child has fever

## Temperature

- Tympanic thermometer
- Paper strips e.g. tempodots (reusable)
- Axillary thermometer → recommended in babies as have small ears

## Tummy

- Note: in boys presenting with abdominal pain ensure to examine testis as **testicular torsion** = **medical emergency**
- Check for masses in groin for males and females as could be **strangulated hernia** (**medical emergency**)
- Bedside test: **urine sample** as e.g. UTI could explain pain

## Vital signs in children

### Resting pulse rate

Age	Bpm
<1 year	110-160
2-5 years	95-140
5-12 years	80-120
> 12 years	60-100

### Capillary refill

- Press of **sternum** or a **digit** at level of heart
- Applying blanching pressure for **5 seconds**
- Measure time for blush to return
- Prolonged cap refill if **>2 seconds**

### Respiratory rate

Age	Normal	Tachypnoea
Neonate	30-50	>60
Infants	20-30	>50
Young children	20-30	>40
Older children	20-30	>30

### Temperature

Normal core temp in a child is usually defined as **36.4 °C**

- < 4 weeks measure in axilla
- > 1 month measured with tympanic thermometer

Fever usually defined as **>37.5 °C**. The very young or those under cancer treatment may not mount a fever response.

## Common childhood presentations

### Coughing. Dx:

#### Acute cough: up to 3-4 weeks

Sudden onset: **foreign body**

Gradual onset – **infection**

- Viral = commonest & self-limiting
- Bacterial
- Asthma with acute exacerbation is often associated with upper respiratory tract infection (URTI)

### Persistent or recurrent cough

- **Asthma**
- Persistent lobar collapse **post pneumonia**
- **Tuberculosis**
- **Gastro-oesophageal reflux**
- Cigarette smoking (passive)
- Following specific respiratory infection e.g. RSV or Pertussis

### Constipation. Dx:

#### Idiopathic with the following contributing factors:

- Fever
- Dehydration
- Dietary and fluid intake
- Psychological issues
- Toilet training difficulties
- Medication side effect
- Family history of constipation

#### Rare underlying conditions including

- **Hirschsprung disease**
- **Hypothyroidism**
- **Coeliac disease**
- Abnormal anorectal anatomy

### Pulling at ear. Dx:

- Infection
- Presence of wax/foreign body in ear
- Teething and referred pain
- Habit e.g. when tired

### Wetting the bed at night

**Primary bedwetting:** *has wet the bed since they were a baby*

#### Without daytime symptoms

- Polyuria
- Bladder dysfunction e.g. small bladder capacity or overactivity
- Sleep arousal difficulties e.g. inability to wake to sensation of full bladder

#### With daytime symptoms

- An overactive bladder
- Urinary tract infection (UTI)
- Chronic constipation
- Structural abnormalities e.g. ectopic ureter
- Neurological disorders

**Secondary bedwetting:** *condition that develops at least 6 months after a person has learned to control his or her bladder*

Causes include:

- Diabetes
- UTI
- Constipation
- psychological problems e.g. emotional or behavioural problems
- family problems (vulnerable child or family)

## Common causes for difficulty in breathing

### Asthma

- Causes **coughing** and **wheezing**
- **Hyperreactive** airways and **mucous production**
- Expiration phase prolonged
- Wheeze can be triggered by: Cigarette smoke, exercise, wheezing, allergies, pollen
- Spacer device easily learned by children
- Small children use facemask
- <1-year-old children do not have typical asthma, so B agonists are less effective

### Croup

- Viral infection of upper airway → airway obstruction and breathing difficulty
- Common in toddlers and present w temperature
- Inflammation of airway causes **barking cough** and **hoarse voice**
- **Stridor**
- Other characteristic signs: intercostal, subcostal, sternal recession and tracheal tug
- Child can get **tired** and can get **respiratory failure** due to obstruction so very important not to upset children with croup

- Responds well to steroids e.g. prednisolone, oral dexamethasone
- **Adrenaline** nebuliser used for more immediate effect for severe croup (doesn't cure but buys time for steroids to work)

### Bronchiolitis

- Mainly affects infancy **1 month - 1 year old**
- SOB, wheeze, mild temp, runny nose
- Caused by few viruses such as **Respiratory syncytial virus**
- Infects lower airways causing secretions so will have wet sounding cough, runny nose etc
- If not feeding, have significant respiratory distress or hypoxic need to admit to hospital for O2 and fluids

### Pneumonia

- Children <3 years old with signs of sepsis
- Chest X-ray often performed as signs are often subtle
- Will seem more lethargic, often refuse food and drink, noisy breathing
- Signs of **respiratory distress**, use **accessory muscles** and **subcostal recession**
- Look out for lethargy, fever and **high HR**

## NICE traffic light system for identifying risk of serious illness:

	Green – low risk	Amber – intermediate risk	Red – high risk
Colour (of skin, lips or tongue)	<ul style="list-style-type: none"> <li>• Normal colour</li> </ul>	<ul style="list-style-type: none"> <li>• Pallor reported by parent/carer</li> </ul>	<ul style="list-style-type: none"> <li>• Pale/mottled/ashen/blue</li> </ul>
Activity	<ul style="list-style-type: none"> <li>• Responds normally to social cues</li> <li>• Content/smiles</li> <li>• Stays awake or awakens quickly</li> <li>• Strong normal cry/not crying</li> </ul>	<ul style="list-style-type: none"> <li>• Not responding normally to social cues</li> <li>• No smile</li> <li>• Wakes only with prolonged stimulation</li> <li>• Decreased activity</li> </ul>	<ul style="list-style-type: none"> <li>• No response to social cues</li> <li>• Appears ill to a healthcare professional</li> <li>• Does not wake or if roused does not stay awake</li> <li>• Weak, high-pitched or continuous cry</li> </ul>
Respiratory		<ul style="list-style-type: none"> <li>• Nasal flaring</li> <li>• Tachypnoea: RR &gt; 50 breaths/minute, age 6–12 months RR &gt; 40 breaths/minute, age &gt; 12 months</li> <li>• Oxygen saturation ≤ 95% in air</li> <li>• Crackles in the chest</li> </ul>	<ul style="list-style-type: none"> <li>• Grunting</li> <li>• Tachypnoea: RR &gt; 60 breaths/minute</li> <li>• Moderate or severe chest indrawing</li> </ul>
Circulation and hydration	<ul style="list-style-type: none"> <li>• Normal skin and eyes</li> <li>• Moist mucous membranes</li> </ul>	<ul style="list-style-type: none"> <li>• Tachycardia: &gt; 160 beats/minute, age &lt; 1 year &gt; 150 beats/minute, age 1–2 years &gt; 140 beats/minute, age 2–5 years</li> <li>• CRT ≥ 3 seconds</li> <li>• Dry mucous membranes</li> <li>• Poor feeding in infants</li> <li>• Reduced urine output</li> </ul>	<ul style="list-style-type: none"> <li>• Reduced skin turgor</li> </ul>
Other	<ul style="list-style-type: none"> <li>• None of the amber or red symptoms or signs</li> </ul>	<ul style="list-style-type: none"> <li>• Age 3–6 months, temperature ≥ 39°C</li> <li>• Fever for ≥ 5 days</li> <li>• Rigors</li> <li>• Swelling of a limb or joint</li> <li>• Non-weight bearing limb/not using an extremity</li> </ul>	<ul style="list-style-type: none"> <li>• Age &lt; 3 months, temperature ≥ 38°C</li> <li>• Non-blanching rash</li> <li>• Bulging fontanelle</li> <li>• Neck stiffness</li> <li>• Status epilepticus</li> <li>• Focal neurological signs</li> <li>• Focal seizures</li> </ul>

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