

Diagnostic Challenges in Paediatrics

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While a diagnosis in a child is frequently straightforward, it can on occasions be complex and difficult. Clearly a diagnosis is more challenging in a non-verbal child. Any condition presents differently across the childhood age groups. While a urinary tract infection presents with dysuria and frequency in a schoolchild, in a baby it frequently presents as a system infection with fever, lethargy, irritability, and a bacteraemia.

A diagnostic error is defined as the failure to establish a timely explanation of the child's medical problem and/or communicate that explanation to the parents. The other part of the error process are the missed opportunities to reach the correct diagnosis.

A number of observers have reported that the diagnostic error rate in adult patients ranges from 5% to 15%. There is less information about the misdiagnosis rates in children. In essence there is a paucity of high quality studies of diagnosis errors in children¹. There needs to be an emphasis on high risk situations, vulnerable groups of children, and the groups of challenging conditions.

The harms associated with misdiagnosis are being increasingly appreciated. The incorrect assessment of a child's symptoms and signs can persist over a prolonged period of time over many weeks and months. The delay can make the treatment of the child less effective or ineffective.

A multistep retrospective chart review reported a 5% error rate. It is likely that the error rates and adverse events are different for children compared with adults. Medication prescribing and administration is more difficult in paediatrics because of the weight based dosing that is required.

A paediatric-specific data base covering the period 1985 to 2005 found that diagnostic errors accounted for one third of medical mishaps in children, with over 90% of cases occurring either at the inpatient or outpatient setting. The major misdiagnoses are predictable. They include meningitis, appendicitis, pneumonia, testicular torsion, fractures, non-accidental injury, congenital heart disease, Kawasaki disease. developmental dysplasia of the hip, and cancers particularly brain tumours. One large study found that in 7.6% cases of appendicitis there was a delay in the diagnosis. Hospitals with lower volumes of cases were more likely to have a misdiagnosis. The diagnosis of testicular torsion is a continuing problem. Delayed presentation, defined as greater than 24 hours from symptom onset, occurred in 6% of cases, the most common misdiagnosis being gastroenteritis. The risk late diagnosis is higher in those children with developmental delay and those with a history of recent genital trauma.

There are delays in 16% of cases of Kawasaki disease beyond day 10, resulting in less effective treatment and a higher risk of coronary aneurysms. Infants under 6 months are at greater risk of misdiagnosis. The diagnosis of brain tumours remains difficult, with just 39% being diagnosed within one month of presentation.

When causation is addressed, cognitive errors and breakdowns in decision making are frequently found. Cognitive errors are due to faulty knowledge, poor history and examination, and poor interpretation of the investigations. A pathway towards an improved diagnostic performance is to provide clinicians with feedback about their decision making. The post-take ward round following the previous night's admissions is a time-honoured, effective teaching tool. The clinical activity in outpatients is more difficult unless there is a debriefing meeting at the conclusion of each clinic.

Quality safety programmes depend heavily on voluntary reporting of diagnostic errors. Reporting is improved by the adoption of non-punitive, systems orientated review processes. Marshall et al² have proposed that we use the term diagnostic learning opportunity (DLO). In their institution the filing of reports increased from 0 to 1.6 per 100 patient admissions.

Communication problems³ between staff members comes up time and again when a significant error occurs. The handover is a critical time. In particular the incoming staff must be alerted to the children in whom a diagnosis has not yet been established and those children whose condition has deteriorated during the day or overnight. Children can exhibit a period of compensatory stabilisation before decompensation. The early signs of deterioration should well flagged so that a timely intervention occurs.

The MPS and the MDU continually emphasise the importance of good note keeping. If there is no written evidence of the care provided to the child, it is more difficult to protect and defend the doctor's professional position. The medical entry should always be timed and dated. It should cover the clinical findings, the tests that have been ordered, the decisions that are made, and the medical plan.

Increasingly, hospitals are switching to electronic records. The advantages include ready availability and legibility. One pitfall to avoid is cutting and pasting the previous day's medical entry. This can lead to error and confusion if the child's condition has deteriorated overnight.

Overdiagnosis is also emerging as a problem in paediatrics. The commonly quoted example is the transient hypoxaemia that is documented in infants recovering from bronchiolitis⁴. It can lead to unnecessary delays in the discharge of clinically stable infants from hospital. Many commentators stress the importance of interpreting the oxygen saturation value in conjunction with the heart rate, respiratory rate, retractions, and the ability to feed satisfactorily. However, the debate about the safe threshold levels remains undecided.

References:

- 1. Marshall TL, Rinke ML, Olson APJ, Brady PW. Diagnostic error in pediatrics: a narrative review. Pediatrics;149:March 2022
- 2. Marshall TL, Ipsaro AJ, Le M. Increasing physician reporting of diagnostic learning opportunities. Pediatrics 2021;147 (1).
- 3. Hoffman JM, Keeling NJ, Forrest CB, Tubbs-Cooley HL, Moore E, Oehler E, Wilson S, Schainker E, Walsh KE. Priorities for pediatric patient safety research. Pediatrics 2022;March.
- 4. Quinonez R, Coon ER, Schroeder AR, Moyer VA. When technology creates uncertainty: pulse oximetry and overdiagnosis of hypoxaemia in bronchiolitis. BMJ 2017;358:Aug 16