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Think Zebras!: When to suspect if your child has primary immunodeficiency disorder (PID) – Dr Asrar Abu Bakar

by [Hidayah](#) • May 31, 2024 • [0 Comments](#)



Infections during childhood is very common. An average healthy child would get at least 4-8 infections per year. Getting these infections are often a normal part of a child's life as the child confronts all the different microorganisms while the child explores the vast world around him or her. That isn't to say that a child should lock him or herself indoors 'free' from all the germs as it is impractical and counter-development to do so. Rather, these 'germs' are an integral part of developing one's immune system so a child can grow normally and socialize with other human beings.

Unfortunately, in a small number of children, their immunity does not develop adequately or they are born with some aspects of their immune system 'missing'. More often, this involves a genetic mutation that has led to their inability to form specific immune cells or molecules within the immune system to fight off infection. This is what we call 'Primary Immunodeficiency' in contrast to 'Secondary Immunodeficiency' such as those occurring due to other chronic illnesses like cancer or drugs that suppresses the immune system which is by far more common.

To date, there are hundreds of Primary Immunodeficiency Disorder (PID) but there could thousands more which we have yet to discover. We know that PID is often difficult to diagnose as we do not routinely perform screening test to look at the genetic make-up of every child born. Furthermore, depending on the individual mutation leading to the specific defect, every condition will present with different symptoms and affects different age groups hence there are many children out there with underlying PID that have yet receive a diagnosis therefore inadequately treated. Unfortunately, many PID conditions carries a debilitating life and may lead to death even at a young age.

A typical infection in a healthy child would often lead to symptoms lasting few days that would either resolve spontaneously or responds quickly to treatment. These children are generally healthy between episodes of infection with normal growth or development. In contrary, children with suspected PID will often be triggered by an infection that is not the typical pathogen we commonly see among children. They can also present with signs and symptoms unusual for a commonly encountered pathogen. These includes an infection that is either too prolonged, too recurrent, too severe and too difficult to treat.

However, there are certain 'themes' and warning signs that would make one suspect a PID disorder. Recognising these signs is important for the child to receive timely diagnosis and appropriate treatment at the age where complications can be minimised and quality of life improved. In some PID conditions, curative therapy such as stem cell transplant is also an option if it's done promptly. Therefore, it's important to have a high index of suspicion in order to screen selected individuals showing these signs.

To make it clearer, let us breakdown few of these warning signs.

1. A child with frequent or recurrent infections

It is rare that for a child who have recovered from an infection, would then go on to re-acquire the same infection in such close proximity of a few days or weeks apart. Often following an infection, a child will develop immunity towards that pathogen that would last months, therefore if the child was to encounter the same pathogen again, the immune system will be able to recognise and destroy the pathogen with immediate effect. Failure to do this could be due to the child not able to develop the necessary immune 'memory' to prevent a subsequent infection from happening again, which is common in certain types of PID. Sometimes, a child might also get recurrence of different infections one after another. Although this can be a normal occurrence in many children especially those with more 'substantial' exposures such as in a crowded nursery or kindergarten, it can also be a sign of PID as they fail to fight off infections easily. Comparing the 'affected' child with another healthy sibling or child sent to the same nursery might be an early clue for many parents although each child have different abilities in fending off infection.

2. A child with prolonged infection

Sometimes certain infections can lead to prolong symptoms requiring the child to visit a doctor or hospital to receive treatment. While the immune system is good at killing off pathogens invading the body, occasionally to speed up the clearance of the pathogen, our immune cells can work in tandem with the help of 'chemicals' called antibiotics or antiviral that can directly destroy the attacking bacteria or viruses. This will help the child recover sooner. However, some children with PID conditions, might not have the immune machinery to work despite the assist from antibiotics as the invading pathogen can 'wall' itself and evade the action of drugs due to lack of functioning immune cells to compensate. Children with these condition often get difficult to treat infections such as abscesses or deep seated infections that don't respond to conventional antibiotics or they might be on antibiotics for many months with little sign of recovery.

3. A child with severe infection

Every so often a child may get severe form of illness from infection that leads to sepsis or meningitis to name a few. These type of infection requires the child to be hospitalised occasionally needing intensive care support. There are many factors that could determine the severity of infection including the type of pathogen, nutritional status, environmental factors and all this can happen even in a child with relatively normal immune function. However the risk of getting severe overwhelming infection increases if your child has an underlying PID condition. This is often true if such severe infection arise when the child has just been born or still an infant but can also occur at any age. One should also be suspicious when a relatively 'benign' bacteria, fungus or viruses leads to a severe form of presentation that would normally cause a mild illness in majority of children.

4. A child with an unusual infection

Infection in children are generally caused by a group of common pathogens circulating in our community among others Influenza, RSV, Rhinovirus, Mycoplasma or Streptococcal Pneumoniae. While some rarer pathogens are often seen in hospital settings causing problems to children with chronic conditions needing prolonged hospitalisations. In children with PID, they can become infected with pathogens that are rarely seen in the community and even without prior visit to hospitals. Another important clue is when following certain live vaccines such as BCG, the child develops infection from the weakened pathogen given in the vaccine itself. Similarly, further investigations should be considered if a child develops an infection that prior vaccine should have helped to prevent. The reason for this is some children with PID do not produce the desired protective antibodies and immune cellular response following vaccination, unlike normal children.

5. A child with another sibling with PID

As mentioned before, most well established PID conditions have a known genetic defect that tends to run in families. Unaffected parents are often carriers of the genetic mutation that could then be passed down to their offsprings. This is common among parents who have biological relationships such as being first or second degree cousins to each other. A sibling diagnosed with PID would warrant further family screening to assess if there are other affected children. However, not many children with underlying PID are diagnosed promptly therefore careful attention need to be made in a history suggestive of an early sibling death usually from an overwhelming or prolong infection.

6. A child with other co-morbidities or immunological conditions

It's also important to look at the overall well-being of the child to look for evidences of a PID disorder. Children with PID often fail to gain adequate weight despite adequate nutritional intake. This could either be due a combination of the underlying condition itself or the extra use of calories to fight off recurrent infections. Due to its close relationship with other immunological disorders including allergies and autoimmune conditions. PID diagnosis should be sought in children with these conditions presenting with multiple or odd infections. Many established syndromic conditions such as Down Syndrome can also have an immunodeficiency component therefore understanding how certain diseases manifest can result in earlier screening and intervention.

It's becoming more important that both physicians and the community become familiar with PID and its rather diverse presentation. Doctors can only suspect when they are given adequate information regarding a child from parents seeking help. To create awareness on the importance of early detection of at-risk children, the Jeffrey Modell Foundation in collaboration with US Centers of Disease Control (CDC) have published a leaflet entitled '10 Warning Signs of Primary Immunodeficiency' which can serve as a good guide to both public readers and professionals. If in doubt, parents are encouraged to speak to their local general practitioner or paediatrician and get referred

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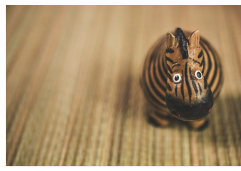


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for early assessment if indicated. Doctors especially those who look after and treat children are also encouraged to be accustomed with these warning signs as it could be a life-changing experience for many parents and children.

There is a saying in the medical world, "when you hear hoof beats, think horses," and trained doctors are taught to focus on the likeliest possibilities when making a diagnosis rather than emphasising on complicated and rare conditions. Common things are common and more often than not tend to follow a common pattern of presentation. Nonetheless, it is when these patterns become too unfamiliar and too off tangent that we need to start discerning on matters other than 'horses'. Whilst PID as an entity if still rare however it is not as uncommon as we generally think as hundreds of children are diagnosed each year allowing them to have access to the care and potentially curative treatment that they need. PID can be a life-long debilitating condition than carries tremendous burden to families and healthcare services if not diagnosed and managed promptly and adequately. So whenever we see a child with weird infections or not dealing with normal infections as expected, we need to think about 'Zebras'!



Written by Asrar Abu Bakar

Paediatrician

Kuliyah of Medicine

International Islamic University Malaysia

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