The Curious Case of a Zebra

During my undergraduate days, I developed an inclination towards pediatrics and decided to pursue my passion in the same speciality. The power of actually becoming competent in treating patients, prescribing drugs and saving tiny lives, gave meaning to 'what a pediatrician can do.'

I was in my first year of pediatric residency. It was my turn to present the new patient in the post-emergency round. I narrated the findings of a year-old child who had severe wasting and stunting with normal routine investigations. I suspected a rare diagnosis and enthusiastically put it across, "Sir, I feel the patient is a case of Seckel syndrome." I have always been very curious about unusual or rare findings in patients. I had the habit of reading further about them.

"Why do you have to make some rare diagnosis?" My lecturer asked me. "You know there is a saying in medicine, 'If you hear hoofbeats, look out for horses and not zebras.' Always think of common diagnoses first and then rarer ones. The majority of cases of wasting and stunting is because of malnutrition, tuberculosis or systemic illnesses like congenital heart disease, renal disease *etc*. Why not attribute the illness to commoner causes?" I replied that the patient had no salient findings in the history and examination related to any of the common causes and in addition had a beak-shaped nose. My senior colleague merely commented that having a different shape did not make it abnormal and we moved on.

The parents were really concerned about why the baby was not growing well, in spite of all the tests being normal. I was uneasy about the diagnosis and felt that further evaluation was required. The patient was counselled about the benign nature of the disease and we discharged him on multivitamins and dietary advice. One year later, the patient was readmitted having received a genetic diagnosis of Seckel syndrome from a geneticist at a premier institute.

I kept wondering about our traditional teaching of seeing a patient as a horse and not as a zebra. We are taught about common diseases and majority of patients are diagnosed correctly. We are also taught to assume that the simplest explanation is usually correct. This is to avoid patients being misdiagnosed with rare illnesses. However, we seem to forget that zebras do exist. If we try to fit every patient into common diagnoses, getting a true diagnosis and treatment can become more difficult for sufferers of rare conditions. We have ample examples of clinical dilemmas like neonatal encephalo-pathy, cerebral palsy, failure to thrive, short stature, recurrent infections etc. in which genetic or metabolic causes may be identified. If we see the patient with unbiased differentials, there is a higher propensity of picking up a rarer diagnosis. We should be taught to consider the rarer differentials of the common presentations so that no child is ever missed.

In making the diagnosis in an individual case, calculations of probability have no meaning. The pertinent question is whether the disease is present or not. Whether it is rare or common does not change the odds in a single patient. The prevalence of a disease in a patient maybe 0.0001 per cent of population, but for the patient, it is 100 % disease. That's why, making efforts to establish the diagnosis are important. The prognosis, treatment options and surveillance depend upon the correct diagnosis. It is practically impossible for each doctor to be updated in each and every aspect of all the known diseases. However, he should be aware of the common symptoms of the uncommon diseases. With this approach, the chances of missing the rarer diseases would be minimized. We can definitely keep our curiosity alive and read more about the uncommon or unusual presentation in a particular patient. We can consult experts. Eventually, this practice will yield lot more gains to the patient who turns out to be a zebra. Since zebras make up a certain percentage of the medical patients; howsoever small, these patients must also be 'thought about'.

A novice resident can definitely be educated about common diseases, and differential diagnoses, but while training on an individual patient, we can at least ignite scientific curiosity. A student can be taught to use the correct, systematic way of how to approach a case. After all, as the saying goes, 'give the people facts and we feed their mind for an hour, awaken curiosity and they feed their own minds for a lifetime'.

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