Disorder of Sex Development: A Case of Missed Opportunity

A 14 year old “boy” born of nonconsanguineous parents presented to us with frequent episodes of vomiting. His illness dated back to the neonatal period when he started having frequent episodes of nonbilious vomiting. Management by local practitioner brought temporary relief. At 4 months of age the parents noticed empty scrotal sacs and the medical advice sought suggested to “wait till 4 years of age.” Abdominal sonography was done at that age, showed intra-abdominal gonads, child was subjected to surgery and the gonads were removed, histology confirmed the gonads to be ovaries. The symptoms continued unabated.

At 12 years of age when the problem increased in severity, an abdominal sonography was done which showed “rudimentary fallopian tubes and uterus and a cervix ending blindly”. An abdominal laparoscopy was done and these structures were removed. Even the second surgery failed to relieve his symptoms.

When this child presented to us, his growth parameters were below third percentile, stretched phallic length was 5.2 cm, both scrotal sacs were empty. There was no hirsuitism. His karyotype was 46 XX and 17OH progesterone level was high (3050 ng/dL), which clinched the diagnosis of congenital adrenal hyperplasia (CAH).

In a newborn phenotypic male with bilateral non-palpable testes, one should consider the possibility of dealing with a virilized female with congenital adrenal hyperplasia, especially if the neonate presents with persistent vomiting(1). In bilateral cryptorchidism with impalpable gonads, anorchia could also be a consideration. An undetectable serum AMH level is a reliable marker when evaluating infants with no palpable gonads(2). This, coupled with elevated serum gonadotropins and an absent testosterone response to hCG stimulation, is predictive of the absence of testes.

A karyotype and 17 OH progesterone estimation could easily have clinched the diagnosis and saved the child unnecessary surgeries. A proper replacement therapy and reconstructive surgery would have allowed the child to be reared up as a normal female.

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Optic Neuritis and Anti tubercular Therapy

I read with interest the recent case report on bilateral optic neuritis due to isoniazid(1), and have the following comments to offer:

The patient described in the case was receiving both isoniazid and streptomycin. The visual side effects of isoniazid have been described in this report but those due to streptomycin have been overlooked. The visual complications described in literature during streptomycin therapy include slowness of accommodation, trembling of distant objects, continuance of image movement on turning the head, disturbance of color vision, papilledema and optic neuritis(2). Though uncommon, visual toxicity of streptomycin should have been kept as a differential diagnosis in their patient.
Paradoxical reactions are commonly seen during treatment of tuberculosis and steroids have been found to be useful in their treatment. Optic neuritis as a paradoxical reaction to tubercular allergen has been reported in adults(3). As the patient reported by them responded to corticosteroids, the visual loss attributed to isoniazid could be an immunologically mediated paradoxical reaction. I also encountered a case where the patient developed optic neuritis on anti-tubercular therapy; she was successfully managed with steroids, and isoniazid was continued.

Thus, the patient described in this case report had a clinical adverse event (optic neuritis) which in addition to isoniazid therapy, could also be explained by concurrent disease (paradoxical reaction) or the other drug (streptomycin). Taking into consideration the above factors, the Naranjo algorithm for adverse drug reaction causality assessment yields a score of 4 (and not 6 as per authors), suggesting that the adverse drug reaction was only possibly (and not probably) related to isoniazid.

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Are These Guidelines Relevant to Indian Situation?

In reference to the recently published review article on oral health in children(1), I have the following comments:

The guidelines presented by the authors, are based primarily on the published policy documents of the American Academy of Pediatric Dentistry (AAPD)(2). The same guidelines may not be applicable to Indian situations. For example, the AAPD document has consistently referred to certain mode of bottle feeding as an important causative factor, and also while suggesting preventive interventions for early childhood caries. In USA, only 43% infants are breastfed at 6 months out of which only 14% are exclusively breastfed; only 23% are receiving breastfeeding at 12 months. Large majority of babies aged 6 months and beyond are bottle fed. In comparison, according to National Family Health Survey-3, the use of bottles with nipples is not common in India. Bottle feeding increases from 5 % under age two months to 18 % at age 9-11 months and declines at older ages. The median duration of breastfeeding is 24.4 months which means breastfeeding remains a predominant mode of feeding the infant and young children in India(3). As the situation of feeding practices is not comparable in both the countries, there is a need to address the guidelines in Indian context.

International and National policies on infant and young child feeding, including IAP’s policy, recommend exclusive breastfeeding for first six months of life followed by introduction of complementary foods after completion of 6 months of age and continued breastfeeding till 2 years or beyond. These policy documents also recommend breastfeeding on demand, day and night. However, Chandna, et al.(1) have given statements and recommendations which are not only contrary to these accepted norms of infant feeding but also without any robust research evidence. The article suggests that breastfeeding on demand is associated...