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.