SPOT DIAGNOSIS (IMAGE GALLERY)

Liver is rarely involved. The incidence of associated anomalies in patients with gastroschisis is relatively infrequent. Intestinal atresia may occur in 10-15 percent of cases {1,4}. Maternal serum alpha- fetoprotein {AFP} is used as screening test although there is a 40 percent rate of false positive results. It is elevated in case of neural tube defects, abdominal wall defects, or atresia of duodenum or oesophagus. Analysis of amniotic AFP and acetylcholinestrase-pseudocholinesterase can be sensitive in detecting abdominal wall defects, especially Gastroschisis. The ultrasonography is helpful beyond 14 weeks {because midgut normally is herniated in 1st trimester} {1}. Unlike with Omphalocele, primary closure is possible in 90 percent patients, but larger defects may require staged repair. Mortality rates have decreased to 5 percent to 10 percent. Enteral feedings may not be established until 2 months after operation {1}.

REFERENCES:

1. Berseth CL, Poenaru D. Abdominal wall problems. In : Taeusch HW, Ballard RA, Gleason CA {eds}. Avery's Diseases of the Newborn. 8th Edn. Elsevier, Saunders, Philadelphia; 2005: 1113-1117.
2. Klein MD. Congenital Abdominal wall Defects. In: Ashcraft KW, Holcomb GW, Murphy JP {eds}. Pediatric Surgery. 4th Edn. Elsevier, Saunders, Philadelphia, 2005: 665-669.
3. Curry JI, McKinney P, Thornton JG, Stringer MD. The aetiology of gastroschisis. BJOG. 2000: 107: 1339-1346.
4. Stringer MD, Sugarman I, Smyth AG. Congenital Defects And Surgical Problems. In: Rennie MJ {editor}. Roberton's Text Book of Neonatology. 4th Edn. Elsevier, Churchill Livingstone, 2005: 733-737.

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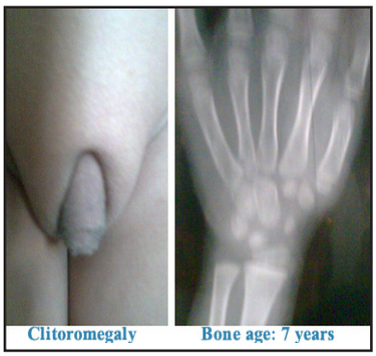

Four Year Old Child with Clitoromegaly

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A four year old female child born from non consanguineous marriage (weight 15 kg and height 102 cm with upper segment, lower segment ratio 1:1) presented with clitoral enlargement in pediatric out-patient department of tertiary teaching care hospital in Haryana. According to parents, problem was present since birth but has increased from past one year. No other physical abnormality was detected in that child. On that basis diagnosis of clitoromegaly was suspected. Blood investigations revealed raised levels of testosterone (1.58 ng, ml), and 17 a-hydroxy progesterone (223.10 ng, ml) with serum electrolytes within normal limits. FSH was raised (3.44 mIU, ml) while LH was normal. Thyroid function tests were normal. Karyotyping revealed female chromosomal complement with no numeric or structural anomalies (46 XX). Magnetic resonance imaging of pelvis showed a small uterus and hypoplastic vagina. No definite testicular tissue was identified. There was prominence of ischiocavernosus and bulbospongiosus muscles forming a penile like structure with urethral meatus at the base of structure. Bilateral adrenal prominence was noted. X-ray wrist revealed increased bone age (7 years).

What is the diagnosis?

Clitoromegaly, otherwise a rare condition, if presents in childhood usually is attributed to congenital adrenal hyperplasia (CAH) in which adrenal gland produces additional androgens and 21 a-hydroxylase deficiency accounts for 95 percent of diagnosed cases. (1) This was a case of simple virilizing CAH with some residual activity of 21-hydroxylase, (2) hence mineralocorticoid deficiency was insignificant and salt wasting did not occur. Adrenal testosterone production was suppressed by glucocorticoid like hydrocortisone.

References

1. Nimkarn S, New MI. Congenital adrenal hyperplasia due to 21-hydroxylase deficiency: A paradigm for prenatal diagnosis and treatment. Ann N Y Acad Sci. 2010; 1192: 5-11
2. Nimkarn S, Lin-Su K, New MI. Steroid 21 hydroxylase deficiency congenital adrenal hyperplasia. Endocrinol Metab Clin North Am. 2009. 38: 699-718

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