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Wernestrup Syndrome or Variant of Vacterl Association – Case Report

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Abstract:- VACTERL association is a useful acronym for a condition characterized by the sporadic, non-random association of specific birth defects on multiple organ systems. It includes vertebral anomalies(V), anal anomalies (A), cardiac anomalies (C), tracheoesophageal fistula(TE), renal anomalies (R), limb defects (L). In addition this, patients may also have other congenital anomalies like external ear malformations, lung lobation defects, intestinal malrotation and genital anomalies. Incidence is estimated approximately 1 in 10000 to 1 in 40000 live births. Ambiguous genitalia may occur with urologic anomaly, described as Richard Wernestrup's syndrome. Richard Wernestrup introduced four cases in 1985 of neonates who were female pseudohermaphroditic with anal imperforation, renal anomalies and other anomalies that were not common to all patients.

I. CASE REPORT

A 10 hours old term, first order male neonate born to non-consanguinously married couples was brought in N.I.C.U of our institute with complaints of abdominal distension and genital anomalies. His weight is 2700gm and delivered by vaginal delivery in Private Hospital. During this pregnancy the mother was registered, underwent regular antenatal check-ups and immunized. She underwent third obstetric scan in her pregnancy which was told to be normal and there was no significant past medical or surgical anomalies or history of family congenital anomalies. On examination baby was alert, vitals – stable. Head to toe examination : mild abdominal distension, ambiguous genitalia, microphallus, hypospadiasis and anal atresia. Systolic murmur better heard on pulmonary area on auscultation.Right kidney palpable on abdominal examination. Other systems normal. The patient was managed in Department of Pediatric Surgery.

II. INVESTIGATIONS

CBC – normal limits. CRP – positive. X-ray shows abdominal distension. USG abdomen : right kidney enlarged in size with loss of corticomedullary differentiation (CMD) and few peripherally arranged cystic lesions - ? dysplastic kidney and polypoidal lesions at the level of perianal region showing minimal vascularity - ? lipoma .2D –echo small PFO,small PDA

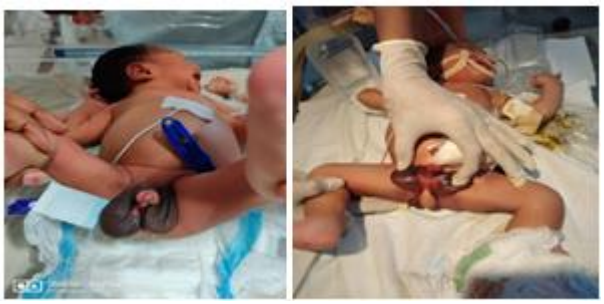

Fig 1: showing ambigus genitalia, hypospadiasis and perianal swellings

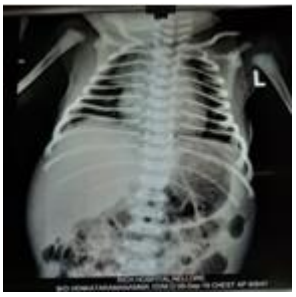

Fig 2: X-ray showing abdominal distension

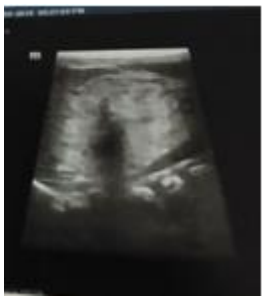

Fig 3: USG abdomen showing loss of CMD

III. TREATMENT

Initially started intravenous fluids, intravenous antibiotics and stage 1 colostomy done and discharged with follow-up after 2 weeks later.

IV. DISCUSSION

The combination of VACTERL abnormalities can present with some known chromosomal abnormalities, including trisomy 13, 18,and 5p- syndrome. VACTERL could be caused by defective Shh (Sonic hedgehog pathway) signalling during human embryogenesis. Polyhydramnios, absence of a gastric bubble, dilated colon, vertebral defects

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and limb abnormalities are certain subtle radiological features that may suggest an affected fetus. The renal and the extrarenal problems may make our case closer to the VACTERL syndrome than the Wernestrup's syndrome. However, tracheoesophageal and vertebral anomalies were not present in our case to be labeled as the VACTERL syndrome. Wernestrup syndrome may additionally have structural defects in heart and skeletal system.

V. CONCLUSION

Association of renal anomalies with imperforated anus and ambiguous genitalia called as wernestrup syndrome.

REFERENCES

[1]. Nelson Textbook of Pediatrics- 21th edition