Volume : 3 | Issue : 7 | July 2014 • ISSN No 2277 - 8179

McKUSICK-KAUFMAN SYNDROME – "A Rare Cause of Neonatal Abdominal Distention"

Medical Science

KEYWORDS :

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McKUSICK-KAUFMAN SYNDROME

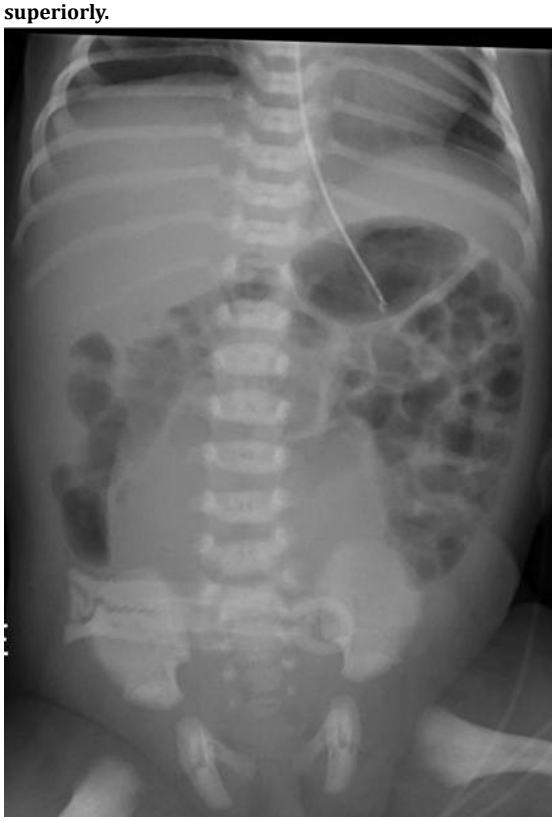
- A newly born female neonate who presented with abdominal distention.
- Baby had passed urine but had not passed stools with normal anal opening.
- Post-axial polydactyly in both hands and in a foot
- Further work up done including xray, usg, ct scan etc.

The results were compared with intra operative findings

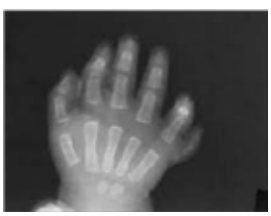
History and Clinical findings

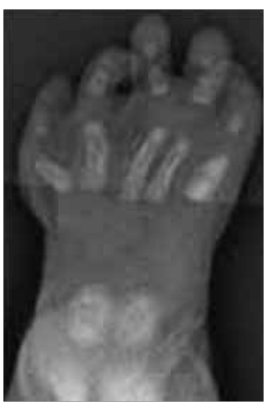
- **Case History:**
- A newly born female neonate who presented with abdominal distention.
- Baby had passed urine but had not passed stools.
- On clinical examination:
  1. Normal anal opening.
  2. Swollen labia majora.
  3. Post-axial polydactyly in both hands and in a foot.

X-ray erect abdomen shows large soft tissue density abdomino-pelvic mass displacing bowel loops laterally and superiorly.

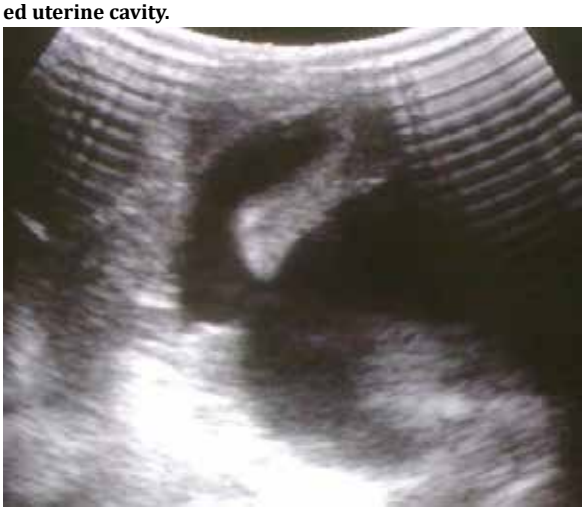


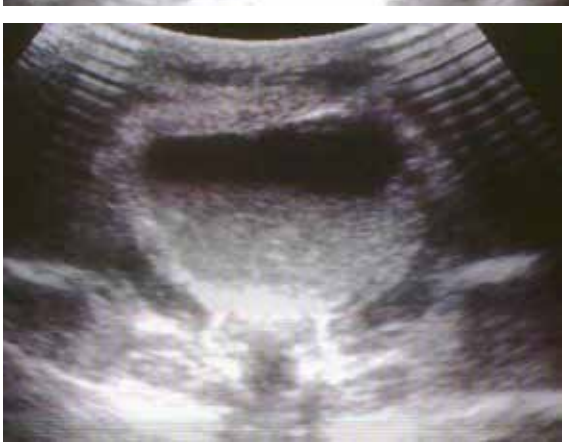
POST AXIAL POLYDACTLY



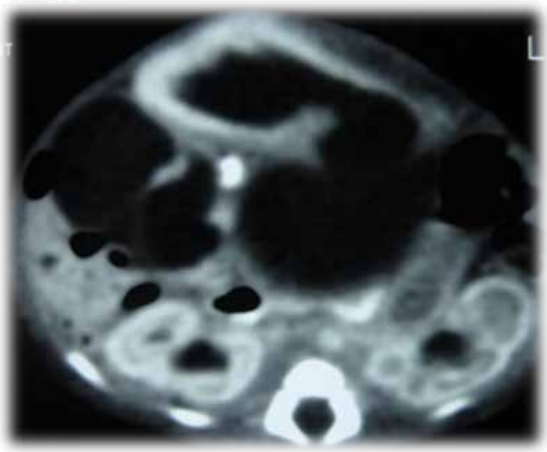


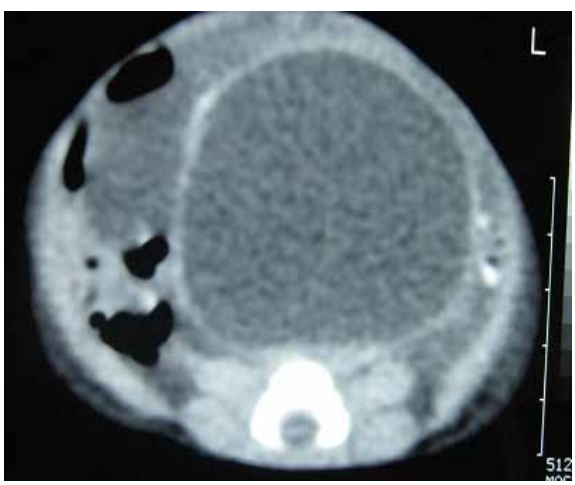
Grey scale USG shows a large hypoechoic cystic lesion in pelvis with fluid –debris level communicating with distended uterine cavity.



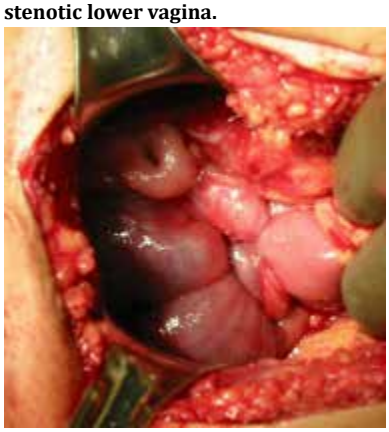


Post contrast CT scan of abdomen and pelvis shows hydrometrocolpos with Bilateral mild hydronephrosis.





Laparatomy confirmed hydrometrocolpos caused due to stenotic lower vagina.



DISSCUSSION:

- An autosomal recessive disease first described by McKusick et al in 1964.
- Cardinal features: HYDROMETROCOLPOS and POLYDACTYLY .
- Also reported as the "hydrometrocolpos-polydactyly syndrome".

- **Causes of hydrometrocolpos:**

1. Vaginal atresia,
2. Imperforate hymen
3. Cervical atresia

which leads to the development of an abdominopelvic mass with regional compression and secondary hydronephrosis.

- Sometimes associated with urogenital sinus anomalies.
- Postaxial polydactyly or syndactyly in 90% of cases.
- Congenital heart defects (atrioventricular canal defect, VSD, hypoplastic left heart) : 10-20% of cases.
- G.I abnormalities (28%) : Imperforate anus, rectovaginal or vesicovaginal fistula, Hirschsprung's disease, and malrotation.

Abnormalities of the eyes (5%) .

- Bardet-Biedl syndrome (BBS) also present with post-axial polydactyly and hydrometrocolpos.
- BBS: characterized by retinal dystrophy or retinitis pigmentosa, postaxial polydactyly, obesity, nephropathy, and mental retardation.
- The diagnosis of BBS can only be made if four of the five major manifestations are present in a person.
- A difficult diagnosis in infancy, as the appearance of several key features is delayed.
- Typically, MKKS is diagnosed in very young children, whereas the diagnosis of BBS often is delayed to the teenage years.
- Hydrometrocolpos should be considered as a possibility in a case of abdominal distension in a newborn female child.

All cases of diagnosed MKKS in infancy should be re-evaluated for retinitis pigmentosa and other signs of BBS as some of these children may be affected by BBS

Differential Diagnosis

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ACKNOWLEDGEMENT:

We take this opportunity to dedicate this work and thanks people who directly contribute to this work and inspired us

- DR.VIPLAV S GANDHI
  PROFFESSOR AND HOD , SMT.SCL HOSPITAL
- DR GURUDATT N THAKKAR
  ASSO.PROFFESSOR, SMT SCL HOSPITAL

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