

7q 34 gene deletion + 14q 34 gene duplication

Signs & Symptoms
Constipation, urine retention,
bowel obstruction, incontinence
low muscle tone, facial asymmetry,
short nose, coarse facial features

Pathophysiology
Currarino
• Sacrum not formed correctly
• can cause urinary + bowel dysfunction/malformation (imperforate anus)
Kleefstra
• Caused by chromosomal deletion
• developmental + intellectual delays

Diagnosics/Labs
genetic testing
X-ray
CT
MRI

Not yet confirmed
Currarino and/or Kleefstra Syndrome

Treatment/Medication
Neurological, psychiatric, + behavioral therapy
Imperforate anus pullthrough (dehiscid)
given colostomy

Nursing Implications
assess bowel sounds
I+O's
proper wound + ostomy care

Patient Teaching
wound + ostomy care
feeding techniques

D.O.B. 2/6/23
Gestational Age 37 Adjusted Gestational Age 46
Birthweight 6 lbs. 1870 oz./ 7870 grams
Current weight 7 lbs. 3240 oz./ 3240 grams
APGAR at birth: N/A born in Midland

Priority Nursing Diagnosis
bowel obstruction / impaired bowel elimination