

distended, palpable bladder. Ultrasound done showed bilateral hydronephrosis and MCU showed a dilated posterior urethra. He underwent valvotomy and circumcision on day 10 of life. Investigations done during neonatal period:

1)ULTRASOUND KUB (PORTABLE): KIDNEYS:Left kidney- Measures 4.4cm, shows dilatation of the calyceal system more than renal pelvis. Ureters are not seen dilated. Right kidney- Measures 4cm, shows dilated pelvicalyceal system with AP diameter of 8mm. Ureters measures: Right proximal and mid ureter-4mm, Distal ureter-4.6mm. Both kidneys appears echogenic with poor corticomedullary differentiation. BLADDER:Empty. No ascites. Impression: -Both kidneys appears echogenic with poor corticomedullary differentiation. -Right hydroureteronephrosis- mild. -Left hydronephrosis- mild (calyces > pelvis).

2)MCU: Prominent posterior urethra with urinary bladder wall irregularities and trabeculations. No vesicoureteric reflux on either sides.

3)NEUROSONOGRAM:Sulci and gyri are normal. Brain parenchyma shows normal echotexture. No evidence of intracerebral/intraventricular bleed. No evidence of hydrocephalus. Corpus callosum is well formed. No evidence of vascular malformation. No extra axial fluid collection. Impression: No evidence of intraventricular hemorrhage/ periventricular leukomalacia.

4)Echocardiogram: PFO Left to Right shunt, Otherwise structurally normal heart, Good LV/RV function, No PAH, Left arch, No coarctation.

5)Karyotyping:47,XY,+21. Interpretation: Chromosome analysis (GTG-banding) revealed an abnormal male chromosome complement with an extra copy of chromosome 21 (Trisomy 21) in all the cells analysed.

He was under multidisciplinary follow-up at AIMS

-Repeat USG (16/12/22)Small sized kidneys with no hydronephrosis / hydroureter or parenchymal thinning. Mild diffuse bladder wall thickening.

-DTPA(08/04/2022)Normally functioning. PUJ not obstructed (function of left kidney :right kidney 56:44%).

-BAEP at 1month of life: Absent V waveform left side.

-OAE repeat at 3months of life:normal.

-He was started on thyronorm at 11months of life.

-He is on regular neurodevelopmental follow-up.

At 6 months and 1yr of life he was admitted with pneumonia and was treated with IV Antibiotics.

PERSONAL HISTORY :

Developmental history:Currently he can sit without support, has bidextrous grasp, says monosyllables, waves bye bye.

Immunization history:Immunized till date as per NIS schedule. 2 doses of influenza vaccine taken

CLINICAL EXAMINATION :

O/E:Dysmorphic facies, febrile

Vitals:Temp-100F, HR-110/min, RR-24/min, BP-94/60mmHg, CRT:<2sec, Spo2-97% in RA.

No pallor, icterus, cyanosis, clubbing, lymphadenopathy, oedema.

Wt-8.8kg(b/w 0 and -2SD), Ht:73cm(At -2 SD), HC-45 cm(b/w -1 and -2 SD)

Mongoloid slant, flat occiput, depressed nasal bridge, open mouth, protruded tongue, webbed neck, single palmar crease, saddle gap.

RS:NVBS, AEBE, B/L crepitations and wheeze.

P/A:Soft, non tender, no organomegaly, BS+

CVS:S1S2 heard normally, no murmurs.

CNS:Conscious, No FND, no signs of meningeal irritation.

INVESTIGATIONS :

Haemogram: