

## Clinical Quiz

### What is the Diagnosis?

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A baby boy was born at 40 weeks and 5 days with birth weight of 3.06 kg by emergency lower segment Cesarean section after a failed induction. Parents were non-consanguineous Chinese and antenatal care was performed in China. Ultrasound scan at 12 weeks showed fetus was small for gestational age and subsequent ultrasound at 24 weeks revealed polyhydramnios.

Upon examination of the newborn, multiple dysmorphic features were noted (Figures 1-8; Figures 1-6 taken at

6 months and Figures 7-8 taken at 10 months). Echocardiogram was remarkable for patent foreman ovale and prominent ascending aorta. Subsequently, bilateral corneal clouding was noted and ophthalmological assessment confirmed bilateral congenital glaucoma for which goniotomy was performed. Upon follow-up, patient displayed severe failure to thrive and global developmental delay and oromotor dysfunction requiring gastrostomy feeding.



Figure 1.



Figure 2.

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N.B. The Editors invite contributions of illustrative clinical cases  
or materials to this section of the journal.



Figure 3.



Figure 4.



Figure 5.



Figure 6.



Figure 7.



Figure 8.