A Case of Emergency Delivery for Non-immune Fetal Hydrops Secondary to Noonan Syndrome: Case Study and Report

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Background
Noonan Syndrome is an autosomal dominant condition, often resulting from de novo mutations. It has a highly variable clinical presentation and prenatal presentations include polyhydramnios, hydrops, and fetal atrial tachycardias.

Aims
We report a case of emergency delivery for fetal hydrops secondary to Noonan Syndrome.

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Case
A 39 year-old, gravida 2 para 1 woman presented to delivery suite at 33+2 gestation with intermittent severe colicky left flank pain, not associated with tightenings. The initial clinical suspicion was for renal stones, though the urinalysis was unremarkable. Her antenatal care up to that point had been uncomplicated with normal fundal heights recorded at antenatal visits. Initial fetal monitoring with cardiotocography was normal. Bedside ultrasound demonstrated new onset of polyhydramnios. She was admitted for observation and monitoring. Routine observations a few hours later identified a foetal tachycardia to 230 beats per minute and repeat bedside ultrasound by senior clinician demonstrated a hydropic fetus with large pericardial and pleural effusions. An emergency lower segment caesarean section was performed, with no maternal complications. Two litres of amniotic fluid drained at delivery. The infant was born with absent tone, distended abdomen and widespread pitting oedema. Neonatal resuscitation included intubation, ascitic tap and decompression of the pleural space, as well as rapid transfer to a level 6 Neonatal Intensive Care Unit.

Results
Trio exome testing demonstrated a de novo pathogenic mutation inf PTPN11 diagnostic of Noonan syndrome. Cardiac abnormalities of the infant were identified post partum including a small ventricular septal defect.

Discussion
This case highlights an example of rapid fetal deterioration in a prenatal presentation of Noonan Syndrome.