Subtle findings at 3rd trimester lead to the antenatal diagnosis of LEOPARD syndrome

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One of the RASopathies: Noonan syndrome, Costello, Cardiofaciocutaneous, Legius syndromes Neurofibromatosis-1

Case:
- Abnormal nuchal translucency 3.2mm
- Normal CMA
- Normal early and late anatomic scans
- Normal echo at 23w
- Normal growth
- Echo 31w– thick septum 5.2mm
- 34w: septum 6mm + dysplastic stenotic pulm. valve= 205cm/sec
- Exome: de-novo missense mutation in PTPN11 gene

• Birth at 37w, 3250gr.
• Facial abnormalities
• AV block 1st degree
• Hypertrophic cardiomyopathy
• Dysplastic & severely stenotic pulmonary valve
• Pulmonary valvuloplasty at 3weeks, high dose β-blockers

Conclusion: A prenatal diagnosis of LEOPARD syndrome is rare; abnormal NT accompanied by cardiac hypertrophy and pulmonary stenosis should raise the suspicion of Noonan/LS and prompt the appropriate consultations. Subtle abnormalities detected early in pregnancy have the potential to become more pronounced over time and therefore warrant periodic evaluation throughout pregnancy.