ABSTRACT# 12

Name of Trainee: Christopher Chow-Parmer (christopher.chow@yale.edu)
Name of Mentor: Robert Elder (robert.elder@yale.edu)
Type of Trainee: Medical Student

Title: Noonan syndrome and congenital heart disease in pregnancy: a case series
CA Chow, KH Campbell, RW Elder. Yale University School of Medicine.

Background: Noonan syndrome is a genetic disorder involving multiple organ systems with a high prevalence of congenital heart defects. Due to increasing rates of genetic testing, as well as improved survival into adulthood, an increasing population of women with Noonan syndrome who wish to become pregnant is emerging. However, scarce data exists regarding the safety and feasibility of pregnancy and delivery in patients with Noonan syndrome, particularly in the context of maternal cardiac disease.

Methods: We performed a retrospective chart review of patients at Yale-New Haven Hospital from 2012-2020 with a diagnosis of Noonan syndrome who had been pregnant. We analyzed medical records for details of their cardiac health as well as the results of their pregnancies, including complications to the mother and/or the fetus/neonate. We analyzed echocardiograms nearest to the time of their pregnancy to quantify pulmonary stenosis or other related congenital cardiac issues.

Results: We identified 5 women with Noonan syndrome who experienced 10 total pregnancies. In terms of maternal cardiac disease, 3/5 patients had mild pulmonary valve stenosis at the time of pregnancy, 2 of which had previously undergone interventional cardiac procedures. 1/5 patients had no cardiac history. Out of 10 pregnancies, 5 (50%) resulted in preterm labor (<37 weeks gestation), with 4 of those 5 being spontaneous preterm labor. 80% of all deliveries were converted to Cesarean section after a trial of labor. 1 pregnancy resulted in intra-uterine fetal demise while 9 pregnancies resulted in the birth of at least 1 live infant, for a total of 10 livebirths (1 set of twins). 6/10 (60%) livebirths required care in the neonatal intensive care unit (NICU).

Conclusions: In our sample of 5 women with Noonan syndrome, the majority of whom had pre-existing though mild heart disease, there were no major maternal complications or long-term morbidity. We observed high rates of prematurity, conversion to Cesarean section, and NICU stays for the neonates. Further study of utero-placental issues in this population may be warranted.

Word Count: 325