Background: Pulmonary vein stenosis (PVS) is a rare and frequently lethal disease. PVS has been reported concomitantly with congenital heart disease (CHD), as well as chronic lung disease (CLD). In this single-center study we examined the association of PVS with CHD and CLD, along with genetic abnormalities seen with PVS.

Methods: An existing registry of all PVS patients seen at Boston Children’s Hospital (BCH) was reviewed, and all patients evaluated between 8/15/2006 to 12/1/2012 were included in this study (n=88). PVS was defined as intraluminal pulmonary venous obstruction in ≥2 vessels with mean pressure gradients >4mm Hg. Thirty-three patients (37.5%) were consented to clinical genetic evaluation and karyotyping.

Results: Eighty-eight patients with PVS (58% male, median age of PVS diagnosis: 5 months, range 0.2-49.6 months) formed our cohort. Twenty-nine patients (33%) were born at < 36 weeks gestation (median: 30 weeks, range: 25-36 weeks). Seventy-five patients (85.2%) had concomitant CHD, of which 17 (19.3%) also had CLD. Six patients (6.8%) were diagnosed with isolated PVS without concomitant CHD or CLD. Fifty-two patients (59.1%) were diagnosed with multiple CHD lesions. Post-natal CHD diagnoses included atrial septal defect (45.5%), anomalous pulmonary venous return (39.8%), patent ductus arteriosus (31.8%), and hypoplastic left heart syndrome (13.6%). Thirty-eight patients (43.1%) were diagnosed with pulmonary hypertension. Karyotyping and clinical genetic evaluation were available in 33 patients (37.5%). Of those, chromosomal abnormalities or specific genetic diagnoses were present in 23 patients (69.7%), while 10 (30.3%) had normal results. Among patients with genetic abnormalities, 6 (26.1%) were diagnosed with trisomy 21, 2 (8.7%) with Smith-Lemli-Opitz syndrome, and 6 (26.1%) with multiple chromosomal abnormalities. No karyotypic abnormalities were highly recurrent.

Conclusion: In patients with PVS, complex CHD was common. Gross chromosomal abnormalities were surprisingly prevalent, but there was significant genetic heterogeneity. Future prospective genetic evaluation in PVS is likely to be informative.