

Cardiac Manifestations in Pearson Syndrome

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INTRODUCTION

- Pearson Syndrome is a rare mitochondrial disease with many multi-system manifestations
- Cardiac manifestations not well studied
- Conduction defects described for other mitochondrial diseases

OBJECTIVES

- Describe the cardiac conduction defects and ventricular dysfunction in this population.
- Describing the timeline of progression of conduction defects

METHODS

- IRB approval
- Patient list obtained from NIH study "Natural History of Pearson Syndrome"
- Inclusion criteria:
Diagnosis of Pearson syndrome
- Exclusion criteria:
Lack of complete data
- Retrospective chart review
- RedCap for data collection

RESULTS

- 12 patients included in study
- Conduction defects were the most common cardiac finding (6/12 patients)
- Most patients (5/6) progressed from BBB to 2nd degree Type 2 AV block and/or CHB
- Progression was over months (mean 9.4months)
- One patient presented with severe symptomatic bradycardia
- All patients with conduction defects had evolution to Kearns Sayre syndrome

Figure 1: Demographics

No. of patients	12
Sex	
Male (n)	8
Female (n)	4
Age at presentation (mean-mnths)	6
Features at presentation (n)	
Sideroblastic anemia	9
Other Cytopenias	4
Metabolic acidosis	2
Corneal edema	1
Transaminitis	1
Jaundice	2

Figure 2: Distribution of cardiac defects

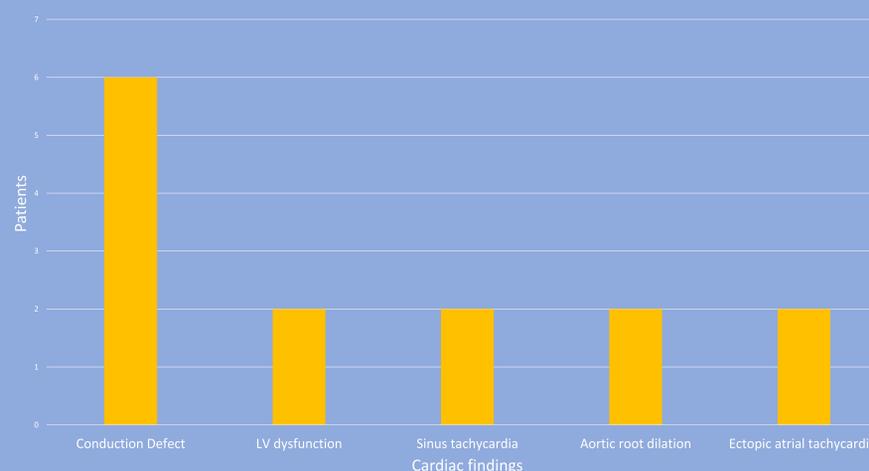


Figure 3: Progression of conduction defect

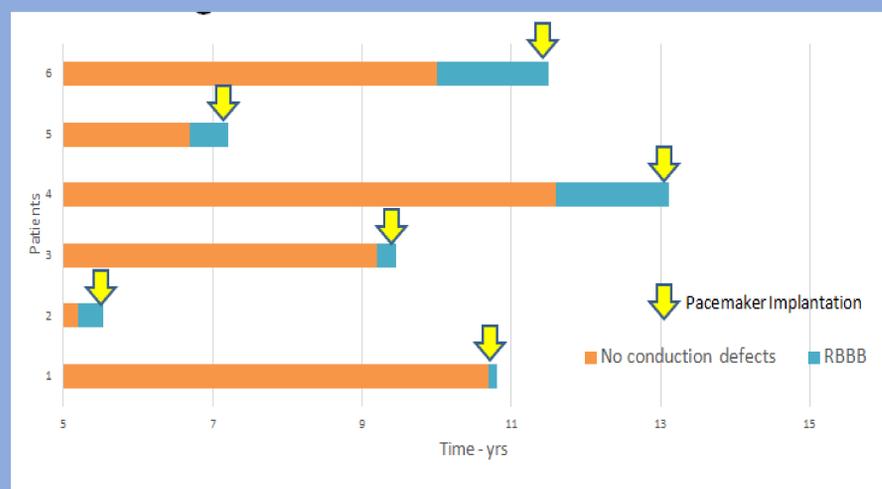
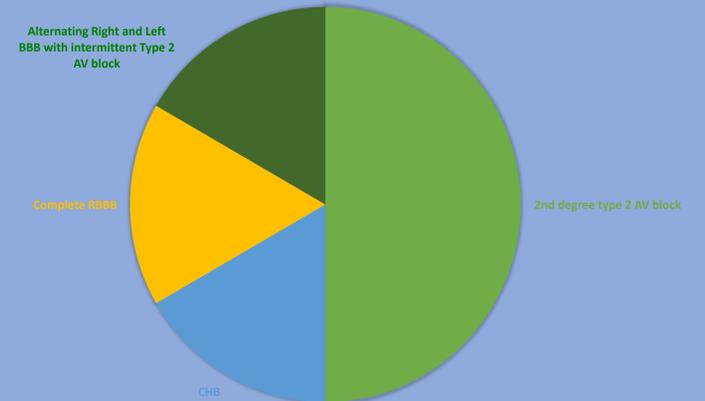


Figure 4: Indications for pacemaker implantation



CONCLUSION

- RBBB may be an indication for pacemaker implantation
- Limitations of the study were that this is retrospective and multicenter.

DISCUSSION

- The current guidelines recommend high grade second degree and complete heart block as class I indications and any evidence of AV node disease (first degree or fascicular block) as class IIb indications for pacemaker implantation in mitochondrial disease patients.
- In our population, our findings suggest that early conduction defects once present, progress quickly and may present with severe symptomatic bradycardia
- Therefore, RBBB may be an indication for pacemaker implantation in this population

REFERENCES

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