Myopia Progression of Pediatric Patients with Stickler Syndrome

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BACKGROUND

- Stickler syndrome is an autosomal dominant connective tissue disorder caused by mutations mainly in COL2A1, COL11A1, and COL11A2 genes.
- A notable ocular manifestation of the disease includes early-onset high myopia (EOHM).
- Treatments are available and have been shown to combat myopia progression in the general population (e.g., atropine), but their effectiveness in genetically induced EOHM is undocumented.
- Previous studies have reported the severity of EOHM in Stickler syndrome patients, but none have performed a longitudinal analysis looking at the rate of progression over time to know whether therapeutic intervention is warranted.

STUDY OBJECTIVE

- To analyze the myopia progression rate in pediatric patients with Stickler syndrome.
  - To compare the estimated annual myopia progression rate at various ages
  - To compare the estimated annual myopia progression rate among patients with COL2A1, COL11A1, and clinically-presumed Stickler syndrome
  - To compare the estimated annual myopia progression rate by laterality

METHODS

- Study Design
  - Retrospective case series
- Setting
  - Large academic tertiary care center
- Population
  - 11 pediatric patients who have clinically or genetically confirmed Stickler syndrome
- Main Outcomes
  - Final refractive error at each visit
  - Estimated annual myopia progression
- Statistical Analysis
  - One-factor ANOVA
  - Student's T-test

RESULTS

Table 1. Estimated average annual progression by age group, mutation, and laterality. One standard deviation is denoted. Statistical significance is calculated using one-factor ANOVA or Student's t-test and is defined at $P < 0.05$.

Parameter	Estimated Annual Myopia Progression	*P*
Age		
6 – 24 months	-1.21D ± 2.36D	0.44
24 – 48 months	0.15D ± 1.87D	
48 – 72 months	0.04D ± 2.39D	
72 – 96 months	0.05D ± 1.63D	
Mutation		
COL2A1	0.05D ± 0.19D	0.89
COL11A1	0.25D ± 0.22D	
Presumed	-0.13D ± 0.08D	
Laterality		
OD	0.22 ± 0.49D	0.26
OS	0.36D ± 0.25D	

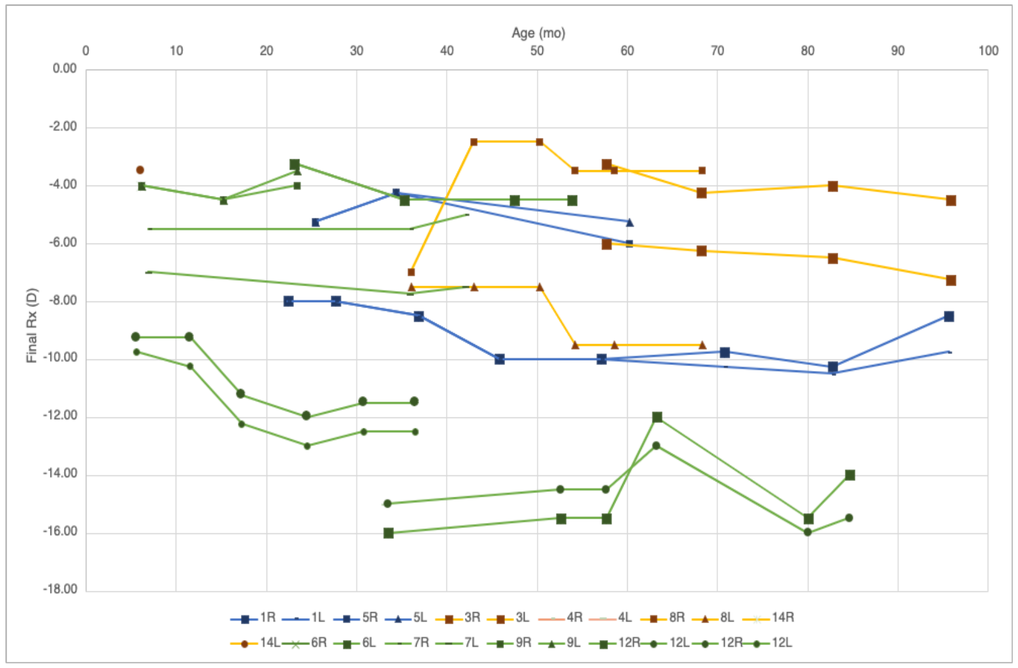

Figure 1. Refractive error of 11 pediatric patients with Sticker Syndrome over time. Green indicate COL2A1 mutation. Yellow indicates COL11A1 mutation. Blue indicates presumed Stickler syndrome.

CONCLUSIONS

- This retrospective case series analysis of 11 pediatric patients with Stickler Syndrome revealed that the estimated annual myopia progression was not significantly different by age, mutation, nor laterality.
- These findings suggest that further research is needed to investigate the efficacy of atropine and other therapeutic interventions in controlling myopia progression in Stickler Syndrome patients.

LIMITATIONS

- This study is limited by its small sample size, retrospective design, and lack of a control group. In addition, the data are only from one center and may not be generalizable to other clinics.
- Also, the rate of myopia progression in Stickler Syndrome patients is highly variable and this study is only able to provide a snapshot of the rate at one point in time.

IMPLICATIONS

- This study provides initial critical insight into whether therapeutic interventions are needed in managing EOHM in pediatric patients with Stickler syndrome.