Complement gene mutations in children with C3 glomerulopathy: Do they affect clinical outcome?

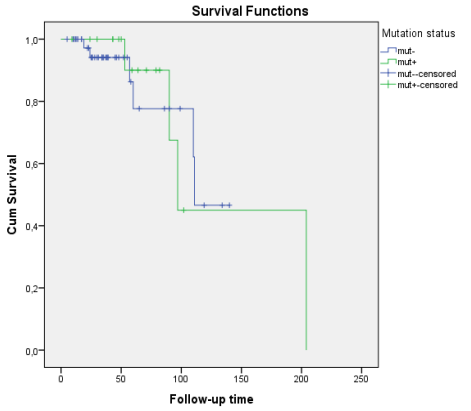
Graphical Abstract

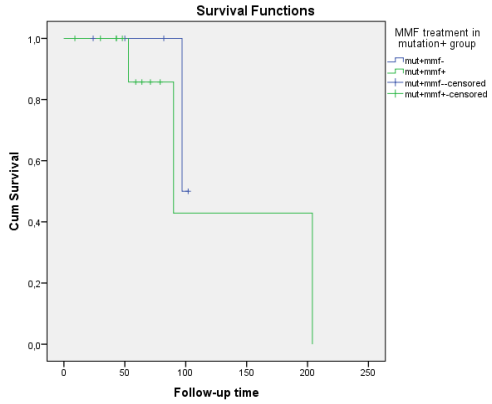
HYPOTHESIS: Having mutations in genes regulating the alternative complement pathway affects kidney survival and response to mycophenolate mofetil therapy in pediatric C3G patients.

DESIGN & OUTCOMES:

Retrospective Study

18 Pediatric Nephrology Clinics → **60 C3GN Patients <18 years old** → **Genetic screening for the alternative complement pathway** → **mutation + n: 43** / **mutation - n: 17**





- The most common mutation in the CH gene (47%)
- While the patients without mutation were most frequently presented with the nephritic syndrome (44.2%), patients with the mutation were most likely to have asymptomatic urinary abnormalities (%47.1, p=0.043).
- Serum parameters and histopathological characteristics were similar between the groups, but hypoalbuminemia was more common in patients without mutation.

Kaplan-Meier analysis for CKD ; Figure1(left): in the group with and without mutation. Mean CKD development time in the group with and without mutation, respectively 139.1±30.5 and 110.4±9.7 months (p:0.98) **Figure2(right):** in the group with mutations by MMF use status. Mean CKD development time in the group with MMF use and don't respectively 133.5±44.9 and 99.5±1.76 months (p:0.572)

CONCLUSION: Patients in the mutation group presented with mild symptoms, were diagnosed relatively late and were not different from the mutation group in terms of MMF treatment response and kidney survival.

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