

## **Antioxidant treatment of Smith-Lemli-Opitz syndrome (SLOS) and retina function determined by electroretinography**

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Smith Lemli Opitz syndrome (SLOS) is an autosomal recessive disease caused by a defect in cholesterol metabolism, resulting in cholesterol deficiency, elevated 7-dehydrocholesterol (7DHC) and progressive retinal degeneration. Retinal function can be monitored by Electroretinograms (ERG's), which measure amplitude, or rod function, and implicit time, the amount of time rods take to respond to stimuli. Cholesterol supplementation and antioxidant supplementation may protect against retinal degeneration in Smith Lemli Opitz Syndrome by decreasing the amount of harmful oxysterols that can damage the retina (Xu). Our study explores the effect of Antioxidant and Cholesterol supplementation on ERG studies in patients with SLOS. We enrolled 8 patients with Smith Lemli-Opitz and provided cholesterol and antioxidant supplements. It should be noted that our population size was limited because SLOS is such a rare disease, but that larger sample sizes would be preferable for future research. Patients were stratified into Mild (n=3), Moderate(n=4), or Severe (n=1) groups based on their baseline serum levels of cholesterol and clinical characteristics. We found that in the mild group, there were overall decreased amplitudes at baseline which improved, and while implicit times remained abnormal, they improved in two of the three patients. In moderate patients, amplitudes tended to improve from moderate to mild deficits and implicit times remained within normal ranges in the majority of patients. In the severe group, there were moderate decreased amplitudes which improved over time, and implicit times remained within a normal range. While this study showed promising correlation between improved retinal function and cholesterol and antioxidant supplementation in patients with SLOS, a randomized control trial with a larger sample size is needed to fully explore this association. Future studies of antioxidants and retinal function may someday help treat other diseases that result in retinal degeneration.