Weighted Pathway Genetic Load Analysis of Hyperbilirubinemic Infants Indicates a Potential Genetic Component for Susceptibility to Bilirubin Neurotoxicity

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Identification of critical pathway genetic load scores related to susceptibility to bilirubin neurotoxicity in neonates is enhanced by weighting genetic variants using CADD scoring.

Results and Conclusions
• Only 1 group comparison in the mPGL+ Tier 1 analysis proved to be statistically significant, High Bilirubin & Mild KSD vs. Low Bilirubin & Severe KSD.
• We hypothesized that increased mPGL scores would correlate with increased susceptibility to bilirubin neurotoxicity but these results show the opposite effect and indicate a need for increased study and thoughtful evaluation of the mPGL score and the gene lists used here.

Next Steps:
• Proceed to a prospective study in an effort to reduce compounding clinical variables and include free bilirubin measurements for data analysis.

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References:
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