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Kernicterus Spectrum Disorders Diagnostic Toolkit: validation using retrospective chart review

Vijaya Dasari

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Research Abstract Title

Submitting/Presenting Author (must be a trainee): Vijaya Dasari (UMKC SOM)
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X Medical Student
☐ **Resident/Psychology Intern (≤ 1 month of dedicated research time)**
☐ **Resident/Ph.D/post graduate (> 1 month of dedicated research time)**
☐ **Fellow**

Primary Mentor (one name only): Dr. Rose Gelineau-Morel
Other authors/contributors involved in project: Dr. Steven Shapiro, Hung-Wen Yeh

IRB Number: STUDY00001051

Describe role of Submitting/Presenting Trainee in this project (limit 150 words):
The medical student submitting/presenting this project, Vijaya Dasari, performed the retrospective chart review by completing the Kernicterus Spectrum Disorders Diagnostic Toolkit for each patient. Statistical analysis was done by CMH statistician Hung-Wen Yeh. Vijaya also helped write the abstract and create the poster presentation and associated tables along with Dr. Gelineau-Morel and Dr. Shapiro.

Background, Objectives/Goal, Methods/Design, Results, Conclusions limited to 500 words

Background:
Kernicterus Spectrum Disorders (KSDs) result from hyperbilirubinemia-induced brain injury. While rare, KSDs can result in devastating neurologic sequelae affecting motor function and tone, causing dystonia and choreoathetosis, and/or auditory neuropathy spectrum disorders. Families of children with undiagnosed neurodevelopmental disorders and a history of neonatal hyperbilirubinemia often question this potential diagnosis. We developed a Toolkit (KSD-TK) questionnaire in an effort to provide a standardized method to predict the likelihood of KSDs.

Objectives/Goal:
This study compares the accuracy of the KSD-TK to clinical diagnoses made by our Children's Mercy Hospital Kernicterus Center of Excellence (KCOE). The primary objective of this study is to determine how accurate the KSD-TK is in predicting a KSD diagnosis. The secondary objective is to determine which risk factors included on the KSD-TK are most predictive of a KSD diagnosis.

Methods/Design:
We retrospectively reviewed charts of 37 patients evaluated between 2011-2019 at the KCOE. We completed a KSD-TK for each patient using data including highest bilirubin, newborn risk factors, neonatal

exam, follow-up exam, auditory testing, presence or absence of enamel dysplasia, and MRI brain results. KSD-TK diagnostic prediction was compared to "gold standard" clinical diagnoses given after KCOE evaluation and used to calculate sensitivity and specificity of the KSD-TK as well as sensitivity, specificity, area under the curve, and accuracy of each risk factor included on the KSD-TK.

Results:

Of the 37 patients, 29 were clinically diagnosed with and 8 without kernicterus. All 14 patients with KSD-TK "definite" kernicterus and 14 of 15 patients with KSD-TK "probable" kernicterus were clinically diagnosed with kernicterus. One of 2 patients with KSD-TK "possible" kernicterus was diagnosed with kernicterus. None of 6 patients with KSD-TK "not kernicterus" were clinically diagnosed with kernicterus. When KSD-TK "definite" and "probable" are combined, the sensitivity of the KSD-TK is 96.6%, while the specificity is 87.5%. Each component of the KSD-TK had a high sensitivity, but only three had a specificity >0.25: auditory testing, follow-up exam, and MRI results.

Conclusions:

The KSD-TK is a promising tool for diagnosing kernicterus with a sensitivity of 96.6% and specificity of 87.5%. Utilizing the KSD-TK as a standardized screening tool could lead to more prompt diagnoses, fewer missed diagnoses, and earlier treatment especially in resource limited settings. Future studies will attempt to replicate these results using KSD-TK's completed by families and referring providers.