PYRUVATE KINASE DEFICIENCY

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The Old Days at Boston Children’s
(There is no) better way to advance the proper cause of medicine than to give our minds to the discovery of the unusual law of Nature by careful investigation of cases of rarer forms of disease. *William Harvey 1657*
Jennifer, Mother, and Bert Glader’s PKD Dogs
(photo used with permission)
Pyruvate Kinase & the Activator

WHY CURE THIS DISORDER?

WHAT IS THE VALUE OF PREVENTING THE COLLECTIVE MEDICAL PROBLEMS OF THESE PATIENTS OVER 30 LIFE-TIMES?
CSC Patients Across Pennsylvania
Big Valley
Pyruvate Kinase as a Medical Problem

Care of the Newborn

Infants with PKD appear to have *erythroblatosis fetalis* with hepatosplenomegally, cutaneous erythropoesis, early and severe indirect hyperbilirubemia.

These infants are at high risk for kernicterus with dystonia and hearing loss.

We recommend in hospital delivery & carrier testing to identify high risk couples.

High intensity phototherapy, intestinal clean-out, early enteral feedings with aggressive hydration.

We use albumin infusions to maintain bilirubin/albumin molar ratio <0.5.

Avoid medications that are known to displace bilirubin from albumin including intralipid.

Transfuse with PRBC to a HCT of 60%.
Pre-Splenectomy Care – Birth to 18 months

Transfuse with washed & irradiated PRBC every 6 weeks to maintain a HCT of 60%, low retic and nucleated RBC counts.

Monitor ferritin & iron and iron binding indices.

Provide childhood immunizations per CDC to complete the 18 month schedule. Measure pneumococcal titers before splenectomy.

Splenectomy at 18-24 months of age.

Stop routine transfusions.

Folic acid 1 mg, methyl-B12 1 mg per day to support increased hematopoiesis.

Amoxicillin 250-500 mg by mouth daily. Bactrim for pen sensitive patients.

Patients should have anti-pneumococcal antibiotics at home and initiate therapy and seek medical evaluation for T>101 F.
The Adult with PKD: Where is your Medical Home?

Folic acid 1 mg, methyl-B12 1 mg per day to support increased hematopoiesis.

Monitor ferritin & iron and iron binding indices - Men become iron toxic earlier than women.

Update immunizations for pneumococcus, Hib vaccine, and meningococcal conjugate vaccine every 5 years. Influenza yearly.

Amoxicillin 500 mg by mouth daily. Bactrim for penicillin sensitive patients.

Patients should have anti-pneumococcal antibiotics at home and initiate therapy and seek medical evaluation for T>101 F.

Monitor ferritin & iron and iron binding indices and adults > 30 should have T2* cardiac & liver iron studies at least every 5 years. Cardiac echo of heart to measure ejection fraction.

Avoid vitamins with iron and foods high in iron – like liver, kidney, and red meats.

Routine echo studies of gall bladder to detect pigment stones.

Pregnancy in affected women may require RBC transfusions because of placental entrapment of PKD cells
Jennifer Cohen & Holmes Morton at the Agios Founder’s Day Meeting
September 20, 2013 (photo used with permission)
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