




## Bertil Glader

Stanford Medicine Professor of Pediatric Hematology/Oncology and Professor, by courtesy, of Pathology

Pediatrics - Hematology & Oncology

 Curriculum Vitae available Online

### CLINICAL OFFICE (PRIMARY)

- **Bass Cancer Center for Childhood Cancer and Blood Diseases**

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### Bio

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### CLINICAL FOCUS

- Pediatric Hematology-Oncology

### ACADEMIC APPOINTMENTS

- Professor, Pediatrics - Hematology & Oncology
- Professor (By courtesy), Pathology
- Member, Maternal & Child Health Research Institute (MCHRI)

### ADMINISTRATIVE APPOINTMENTS

- Instructor in Pediatrics, Harvard Medical School, (1973-1974)
- Assistant Professor of Pediatrics, Harvard Medical School, (1974-1977)
- Associate Professor of Pediatrics, Stanford University, (1977-1987)
- Professor of Pediatrics, Stanford University, (1987- present)

### PROFESSIONAL EDUCATION

- Internship: Stanford Health Care at Lucile Packard Children's Hospital (1969) CA
- Fellowship: Boston Childrens Hospital Pediatric Hematology and Oncology Fellowship (1974) MA
- Residency: Children's Hospital Boston Medical Center Pediatric Residency (1973) MA
- Medical Education: Northwestern University Feinberg School of Medicine (1968) IL
- Board Certification: Pediatrics, American Board of Pediatrics (1982)
- Board Certification: Pediatric Hematology-Oncology, American Board of Pediatrics (1982)
- Board Certification: Hematology, American Board of Pathology (1983)
- BA, Northwestern University , Philosophy (1961)

- PhD, University of Illinois , Physiology (1967)
- MD, Northwestern University , Medicine (1968)

## Research & Scholarship

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### CURRENT RESEARCH AND SCHOLARLY INTERESTS

Hematology/Oncology, biology, and treatment of bone marrow failure disorders, hereditary red blood cell disorders-clinical trials.

### CLINICAL TRIALS

- A Study of AG-348 in Adult Participants With Pyruvate Kinase (PK) Deficiency, Recruiting
- Pyruvate Kinase Deficiency Natural History Study, Not Recruiting

## Publications

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### PUBLICATIONS

- **Diagnosis and management of pyruvate kinase deficiency: international expert guidelines.** *The Lancet. Haematology*  
Al-Samkari, H., Shehata, N., Lang-Robertson, K., Bianchi, P., Glenthøj, A., Sheth, S., Neufeld, E. J., Rees, D. C., Chonat, S., Kuo, K. H., Rothman, J. A., Barcellini, W., van Beers, et al  
2024
- **LENTIVIRAL-MEDIATED GENE THERAPY FOR SEVERE PYRUVATE KINASE DEFICIENCY: RESULTS FROM AN ONGOING GLOBAL PHASE 1 STUDY**  
Lopez Lorenzo, J., Shah, A., Sevilla, J., Navarro, S., Llanos, L., de Camino Gaisse, B., Sanchez, S., Zubicaray, J., Glader, B., Chien, M., Quintana Bustamante, O., Zeini, M., Choi, et al  
SPRINGER NATURE.2023: 275-276
- **LENTIVIRAL-MEDIATED GENE THERAPY FOR SEVERE PYRUVATE KINASE DEFICIENCY: GLOBAL PHASE 1 STUDY RESULTS**  
Shah, A., Lorenzo, J., Sevilla, J., Navarro, S., Llanos, L., Gaisse, B., Sanchez, S., Zubicaray, J., Glader, B., Chien, M., Bustamante, O. Q., Zeini, M., Choi, et al  
WILEY.2023: S133-S134
- **Global Phase 1 Study Results of Lentiviral Mediated Gene Therapy for Severe Pyruvate Kinase Deficiency**  
Shah, A. J., Lopez Lorenzo, J., Sevilla, J., Navarro, S., Llanos, L., de Camino Gaisse, B., Sanchez, S., Zubicaray, J., Glader, B., Chien, M., Quintana Bustamante, O., Zeini, M., Choi, et al  
CELL PRESS.2023: 118-119
- **SATB1 chromatin loops regulate Megakaryocyte/Erythroid Progenitor Expansion by facilitating HSP70 and GATA1 induction.** *Stem cells (Dayton, Ohio)*  
Wilkes, M. C., Chae, H. D., Scanlon, V., Cepika, A. M., Wentworth, E. P., Saxena, M., Eskin, A., Chen, Z., Glader, B., Roncarolo, M. G., Nelson, S. F., Sakamoto, K. M.  
2023
- **The Pyruvate Kinase Deficiency Global Longitudinal (Peak) Registry: rationale and study design.** *BMJ open*  
Grace, R. F., van Beers, E. J., Vives Corrons, J. L., Glader, B., Glenthøj, A., Kanno, H., Kuo, K. H., Lander, C., Layton, D. M., Pospíšilová, D., Viprakasit, V., Li, J., Yan, et al  
2023; 13 (3): e063605
- **Case report: Refractory Evans syndrome in two patients with spondyloenchondrodysplasia with immune dysregulation treated successfully with JAK1/JAK2 inhibition.** *Frontiers in immunology*  
Gernez, Y., Narula, M., Cepika, A., Valdes Camacho, J., Hoyte, E. G., Mouradian, K., Glader, B., Singh, D., Sathi, B., Rao, L., Tolin, A. L., Weinberg, K. I., Lewis, et al  
2023; 14: 1328005
- **Interim Results from an Ongoing Global Phase 1 Study of Lentiviral Mediated Gene Therapy for Pyruvate Kinase Deficiency**  
Lopez Lorenzo, J. L., Shah, A. J., Sevilla, J., Navarro, S., Llanos, L., de Camino Gaisse, B., Sanchez, S., Zubicaray, J., Glader, B., Chien, M., Bustamante, O., Zeini, M., Choi, et al  
MARY ANN LIEBERT, INC.2022: A58
- **Animal models of Diamond-Blackfan anemia: updates and challenges.** *Haematologica*

Liu, Y. L., Shibuya, A., Glader, B., Wilkes, M. C., Barna, M., Sakamoto, K. M.  
2022

- **Comorbidities and Complications in Pediatric Patients with Pyruvate Kinase Deficiency Enrolled in the Peak Registry**  
Grace, R. F., Glenthøj, A., Lander, C., van Beers, E. J., Kanno, H., Corrons, J., Bianchi, P., Pospisilova, D., Williams, J., Yan, Y., McGee, B., Glader, B.  
AMER SOC HEMATOLOGY.2022: 5316-5318
- **The Launch of Two Sub-Studies of the Peak Registry, a Global, Longitudinal Study of Pyruvate Kinase Deficiency**  
Lander, C., Grace, R. F., van Beers, E. J., Vives Corrons, J., Glader, B., Kuo, K. M., Glenthøj, A., Pospisilova, D., Layton, D., Boscoe, A., Smith, S., McGee, B., Bianchi, et al  
AMER SOC HEMATOLOGY.2022: 11041-11042
- **Age of Onset of Complications in Patients with Pyruvate Kinase Deficiency: Analysis from the Peak Registry**  
Glenthøj, A., Grace, R. F., van Beers, E. J., Vives Corrons, J., Glader, B., Kuo, K. M., Lander, C., Pospisilova, D., Kanno, H., Williams, J., Yan, Y., McGee, B., Bianchi, et al  
AMER SOC HEMATOLOGY.2022: 5323-5325
- **Accurate Identification of Hemoglobin Variants By Top-Down Protein Analysis Using Capillary Electrophoresis-HighResolution Mass Spectrometry**  
Luo, R., Wong, C., Xia, J., Glader, B., Shi, R., Zehnder, J. L.  
AMER SOC HEMATOLOGY.2022: 5384-5386
- **Lentiviral-mediated Gene Therapy for Adults and Children with Severe Pyruvate Kinase Deficiency: Results from an Ongoing Global Phase 1 Study**  
Shah, A. J., Lopez Lorenzo, J., Sevilla, J., Navarro, S., Llanos, L., de Camino Gaisse, B., Sanchez, S., Zubicaray, J., Glader, B., Chien, M., Bustamante, O., Zeini, M., Choi, et al  
AMER SOC HEMATOLOGY.2022: 4902-4903
- **Neutral-Coating Capillary Electrophoresis Coupled with High-Resolution Mass Spectrometry for Top-Down Identification of Hemoglobin Variants.** *Clinical chemistry*  
Luo, R. Y., Wong, C., Xia, J. Q., Glader, B. E., Shi, R., Zehnder, J. L.  
2022
- **Changing the Treatment Paradigm for Pyruvate Kinase Deficiency with Lentiviral Mediated Gene Therapy: Interim Results from an Ongoing Global Phase 1 Study**  
Shah, A. J., Lopez Lorenzo, J. L., Navarro, S., Sevilla, J., Llanos, L., de Camino Gaisse, B., Sanchez, S., Glader, B., Chien, M., Quintana Bustamante, O., Zeini, M., Choi, G., Nicoletti, et al  
CELL PRESS.2022: 170
- **Who Should be Eligible for Gene Therapy Clinical Trials in Red Blood Cell Pyruvate Kinase Deficiency (PKD)?: Towards an Expanded Definition of Severe PKD.** *American journal of hematology*  
Schwartz, J. D., Barcellini, W., Grace, R. F., Bianchi, P., Zanella, A., Lopez Lorenzo, J. L., Sevilla, J., Shah, A. J., Glader, B., Nicoletti, E., Ordonez, S. N., Segovia, J. C.  
1800
- **A Congenital Anemia Reveals Distinct Targeting Mechanisms for Master Transcription Factor GATA1.** *Blood*  
Ludwig, L., Lareau, C. A., Bao, E. L., Liu, N., Utsugisawa, T., Tseng, A. M., Myers, S. A., Verboon, J. M., Ulirsch, J. C., Luo, W., Muus, C., Fiorini, C., Olive, et al  
2022
- **Downregulation of SATB1 by miRNAs Reduces Megakaryocyte/Erythroid Progenitor Expansion in pre-clinical models of Diamond Blackfan Anemia** *Experimental Hematology*  
Wilkes, M. C., Scanlon, V., Shibuya, A., Cepika, A., Eskin, A., Chen, Z., Narla, A., Glader, B., Roncarolo, M., Nelson, S. F., Sakamoto, K. M.  
2022
- **Characterizing Iron Overload By Age in Patients Diagnosed with Pyruvate Kinase Deficiency - a Descriptive Analysis from the Peak Registry**  
Bianchi, P., Grace, R. F., Vives Corrons, J., Glader, B., Glenthøj, A., Kanno, H., Kuo, K. M., Lander, C., Layton, D., Pospisilova, D., Viprakasit, V., Williams, J., Yan, et al  
AMER SOC HEMATOLOGY.2021
- **Lentiviral Mediated Gene Therapy for Pyruvate Kinase Deficiency: Interim Results of a Global Phase 1 Study for Adult and Pediatric Patients**  
Shah, A. J., Lopez Lorenzo, J., Navarro, S., Sevilla, J., Llanos, L., Perez de Camino Gaisse, B., Sanchez, S., Glader, B., Chien, M., Quintana Bustamante, O., Beard, B. C., Law, K. M., Zeini, et al

AMER SOC HEMATOLOGY.2021

- **Nutritional Supplements, Ginseng and Leucine, Increase Erythropoiesis in Diamond Blackfan Anemia Models through Inhibition of Nemo-like Kinase**  
Wilkes, M. C., Shibuya, A., Mercado, J. D., Narla, A., Glader, B., Sakamoto, K. M.  
AMER SOC HEMATOLOGY.2021
- **The Serine Threonine Kinase Inhibitor Ots-167 Improves Erythropoiesis through Suppression of Nlk Activity in Diamond Blackfan Anemia Models**  
Shibuya, A., Wilkes, M. C., Serrano, M., Flygare, J., Glader, B., Narla, A., Sakamoto, K. M.  
AMER SOC HEMATOLOGY.2021
- **BASELINE CHARACTERISTICS BY AGE OF A GLOBAL COHORT OF PATIENTS DIAGNOSED WITH PYRUVATE KINASE DEFICIENCY - A DESCRIPTIVE ANALYSIS FROM THE PEAK REGISTRY**  
Bianchi, P., van Beers, E. J., Vives Corrons, J., Glader, B., Glenthøj, A., Kanno, H., Kuo, K. M., Lander, C., Layton, D. M., Pospisilova, D., Viprakasit, V., Williams, J., Yan, et al  
FERRATA STORTI FOUNDATION.2021: 107-108
- **Lentiviral mediated gene therapy for pyruvate kinase deficiency: Interim results of a global phase 1 study for adult and pediatric patients**  
Lorenzo, J., Shah, A. J., Navarro, S., Sevilla, J., Llanos, L., Gaisse, B., Sanchez, S., Glader, B., Chien, M., Bustamante, O., Beard, B. C., Zeini, M., Choi, et al  
MARY ANN LIEBERT, INC.2021: A25
- **Lentiviral mediated gene therapy for pyruvate kinase deficiency: Interim results of a global phase 1 study for adult and pediatric patients**  
Lopez Lorenzo, J. L., Shah, A. J., Navarro, S., Sevilla, J., Llanos, L., de Camino Gaisse, B. P., Sanchez, S., Glader, B., Chien, M., Quintana Bustamante, O., Beard, B. C., Zeini, M., Choi, et al  
MARY ANN LIEBERT, INC.2021: A25
- **GINSENOSE RB1 AND METFORMIN IMPROVES ERYTHROPOIESIS IN MODELS OF DIAMOND BLACKFAN ANEMIA BY TARGETING NEMO-LIKE KINASE**  
Wilkes, M., Jung, K., Lee, B., Saxena, M., Sathianathen, R., Mercado, J., Perez, C., Flygare, J., Narla, A., Glader, B., Sakamoto, K.  
ELSEVIER SCIENCE INC.2021: S110
- **SLC25A38 Congenital Sideroblastic Anemia: Phenotypes and genotypes of 31 individuals from 24 families, including 11 novel mutations, and a review of the literature. *Human mutation***  
Heeney, M. M., Berhe, S., Campagna, D. R., Oved, J. H., Kurre, P., Shaw, P. J., Teo, J., Shanap, M. A., Hassab, H. M., Glader, B. E., Shah, S., Yoshimi, A., Ameri, et al  
2021
- **The active component of Ginseng, Ginsenoside Rb1, improves erythropoiesis in models of Diamond Blackfan Anemia by targeting Nemo-like Kinase. *The Journal of biological chemistry***  
Wilkes, M. C., Jung, K., Lee, B. E., Saxena, M., Sathianathen, R. S., Mercado, J. D., Perez, C., Flygare, J., Narla, A., Glader, B., Sakamoto, K. M.  
2021: 100988
- **Pyruvate kinase deficiency in children. *Pediatric blood & cancer***  
Chonat, S., Eber, S. W., Holzhauser, S., Kollmar, N., Morton, D. H., Glader, B., Neufeld, E. J., Yaish, H. M., Rothman, J. A., Sharma, M., Ravindranath, Y., Wang, H., Breakey, et al  
2021: e29148
- **Lentiviral Mediated Gene Therapy for Pyruvate Kinase Deficiency: Updated Results of a Global Phase 1 Study for Adult and Pediatric Patients**  
Lopez Lorenzo, J., Shah, A. J., Navarro, S., Sevilla, J., Llanos, L., Perez Camino de Gaisse, B., Sanchez, S., Glader, B., Chien, M., Quintana-Bustamante, O., Beard, B. C., Law, K. M., Zeini, et al  
CELL PRESS.2021: 42-43
- **Metformin-induced suppression of NLK improves erythropoiesis in pre-clinical models of Diamond Blackfan Anemia through induction of miR-26a. *Experimental hematology***  
Wilkes, M. C., Siva, K., Varetti, G., Mercado, J., Wentworth, E. P., Perez, C., Saxena, M., Kam, S., Kapur, S., Chen, J., Narla, A., Glader, B., Lin, et al  
2020
- **Diamond Blackfan anemia is mediated by hyperactive Nemo-like kinase. *Nature communications***  
Wilkes, M. C., Siva, K., Chen, J., Varetti, G., Youn, M. Y., Chae, H., Ek, F., Olsson, R., Lundback, T., Dever, D. P., Nishimura, T., Narla, A., Glader, et al  
2020; 11 (1): 3344
- **The variable manifestations of disease in pyruvate kinase deficiency and their management. *Haematologica***  
Al-Samkari, H., van Beers, E. J., Kuo, K. H., Barcellini, W., Bianchi, P., Glenthøj, A. B., Manu-Pereira, M., van Wijk, R., Glader, B., Grace, R. F.

2020

- **Genotype-Phenotype Correlation and Molecular Heterogeneity in Pyruvate Kinase Deficiency.** *American journal of hematology*  
Bianchi, P., Fermo, E., Lezon-Geyda, K., van Beers, E., Morton, D. H., Barcellini, W., Glader, B., Chonat, S., Ravindranath, Y., Newburger, P., Kollmar, N., Despotovic, J., Verhovsek, et al  
2020
- **The pyruvate kinase (PK) to hexokinase enzyme activity ratio and erythrocyte PK protein level in the diagnosis and phenotype of PK deficiency.** *British journal of haematology*  
Al-Samkari, H. n., Addonizio, K. n., Glader, B. n., Morton, D. H., Chonat, S. n., Thompson, A. A., Kuo, K. H., Ravindranath, Y. n., Wang, H. n., Rothman, J. A., Kwiatkowski, J. L., Kung, C. n., Kosinski, et al  
2020
- **The variable manifestations of disease in pyruvate kinase deficiency and their management.** *Haematologica*  
Al-Samkari, H. n., Van Beers, E. J., Kuo, K. H., Barcellini, W. n., Bianchi, P. n., Glenthøj, A. n., Del Mar Mañú Pereira, M. n., Van Wijk, R. n., Glader, B. n., Grace, R. F.  
2020; 105 (9): 2229–39
- **Comorbidities and complications in adults with pyruvate kinase deficiency.** *European journal of haematology*  
Boscoe, A. N., Yan, Y. n., Hedgeman, E. n., van Beers, E. J., Al-Samkari, H. n., Barcellini, W. n., Eber, S. W., Glader, B. n., Yaish, H. M., Chonat, S. n., Sharma, M. n., Kuo, K. H., Neufeld, et al  
2020
- **Characterization of the Severe Phenotype of Pyruvate Kinase Deficiency.** *American journal of hematology*  
Al-Samkari, H. n., van Beers, E. J., Morton, D. H., Barcellini, W. n., Eber, S. W., Glader, B. n., Yaish, H. M., Chonat, S. n., Kuo, K. H., Kollmar, N. n., Despotovic, J. M., Pospíšilová, D. n., Knoll, et al  
2020
- **L-leucine improves anemia and growth in patients with transfusion-dependent Diamond-Blackfan anemia: Results from a multicenter pilot phase I/II study from the Diamond-Blackfan Anemia Registry.** *Pediatric blood & cancer*  
Vlachos, A. n., Atsidaftos, E. n., Lababidi, M. L., Muir, E. n., Rogers, Z. R., Alhushki, W. n., Bernstein, J. n., Glader, B. n., Gruner, B. n., Hartung, H. n., Knoll, C. n., Loew, T. n., Nalepa, et al  
2020: e28748
- **Pharmacological Inhibition of Nemo-like Kinase Rescues mTOR-Mediated Translation and Primes Progenitors for Leucine-Stimulated Erythroid Expansion in Pre-Clinical Models of Diamond Blackfan Anemia**  
Wilkes, M. C., Mercado, J. D., Saxena, M., Chen, J., Siva, K., Chae, H., Youn, M., Wentworth, E., Jung, K., Sathianathen, R., Patel, H., Glader, B., Narla, et al  
AMER SOC HEMATOLOGY.2019
- **Long-Term Safety and Efficacy of Mitapivat (AG-348), a Pyruvate Kinase Activator, in Patients with Pyruvate Kinase Deficiency: The DRIVE PK Study**  
Grace, R. F., Layton, D., Galacteros, F., Barcellini, W., van Beers, E. J., Yaish, H. M., Ravindranath, Y., Kuo, K. M., Sheth, S., Kwiatkowski, J. L., Hua, L., Hawkins, P. F., Mix, et al  
AMER SOC HEMATOLOGY.2019
- **An Ongoing Global, Longitudinal, Observational Study of Patients with Pyruvate Kinase Deficiency: The PEAK Registry**  
Grace, R. F., Bianchi, P., Glader, B., Glenthøj, A., Jones, B., Kanno, H., Kuo, K. M., Layton, D., van Beers, E. J., Corrons, J., Xu, T.  
AMER SOC HEMATOLOGY.2019
- **Pyruvate Kinase (PK) Protein and Enzyme Levels in the Diagnosis and Clinical Phenotype of PK Deficiency**  
Addonizio, K., Al-Samkari, H., Glader, B., Morton, D., Chonat, S., Thompson, A. A., Kuo, K. M., Ravindranath, Y., Wang, H., Rothman, J. A., Kwiatkowski, J. L., Kung, C., Kosinski, et al  
AMER SOC HEMATOLOGY.2019
- **Comorbidities and Complications in Adults with Pyruvate Kinase Deficiency**  
Boscoe, A. N., Yan, Y., Hedgeman, E., van Beers, E. J., Al-Samkari, H., Barcellini, W., Eber, S. W., Glader, B., Yaish, H. M., Chonat, S., Rothman, J. A., Sharma, M., Kollmar, et al  
AMER SOC HEMATOLOGY.2019
- **Characterization of the Severe Phenotype of Pyruvate Kinase Deficiency**  
Al-Samkari, H., Van Beers, E. J., Morton, D., Barcellini, W., Eber, S. W., Glader, B., Yaish, H. M., Chonat, S., Kuo, K. M., Kollmar, N., Despotovic, J. M., Pospíšilová, D., Knoll, et al  
AMER SOC HEMATOLOGY.2019

- **Immunosuppressive therapy for pediatric aplastic anemia: a North American Pediatric Aplastic Anemia Consortium study** *HAEMATOLOGICA*  
Rogers, Z. R., Nakano, T. A., Olson, T. S., Bertuch, A. A., Wang, W., Gillio, A., Coates, T. D., Chawla, A., Castillo, P., Kurre, P., Gamper, C., Bennett, C. M., Joshi, et al  
2019; 104 (10): 1974–83
- **MMP9 inhibition increases erythropoiesis in RPS14-deficient del(5q) MDS models through suppression of TGF-beta pathways.** *Blood advances*  
Youn, M., Huang, H., Chen, C., Kam, S., Wilkes, M. C., Chae, H., Sridhar, K. J., Greenberg, P. L., Glader, B., Narla, A., Lin, S., Sakamoto, K. M.  
2019; 3 (18): 2751–63
- **INHIBITION OF NEMO-LIKE KINASE IMPROVES ERYTHROPOIESIS IN MODELS OF DIAMOND BLACKFAN ANEMIA**  
Takasaki, K., Wilkes, M., Chen, J., Siva, K., Varetta, G., Dever, D., Youn, M., Chae, H., Mercado, J., Saxena, M., Narla, A., Glader, B., Porteus, et al  
WILEY.2019
- **Immunosuppressive therapy for pediatric aplastic anemia: a North American Pediatric Aplastic Anemia Consortium study.** *Haematologica*  
Rogers, Z. R., Nakano, T. A., Olson, T. S., Bertuch, A. A., Wang, W., Gillio, A., Coates, T. D., Chawla, A., Castillo, P., Kurre, P., Gamper, C., Bennett, C. M., Joshi, et al  
2019
- **Prevalence and management of iron overload in pyruvate kinase deficiency: report from the Pyruvate Kinase Deficiency Natural History Study** *HAEMATOLOGICA*  
van Beers, E. J., van Straaten, S., Morton, D., Barcellini, W., Eber, S. W., Glader, B., Yaish, H. M., Chonat, S., Kwiatkowski, J. L., Rothman, J. A., Sharma, M., Neufeld, E. J., Sheth, et al  
2019; 104 (2): E51–E53
- **Addressing the diagnostic gaps in pyruvate kinase deficiency: Consensus recommendations on the diagnosis of pyruvate kinase deficiency** *AMERICAN JOURNAL OF HEMATOLOGY*  
Bianchi, P., Fermo, E., Glader, B., Kanno, H., Agarwal, A., Barcellini, W., Eber, S., Hoyer, J. D., Kuter, D. J., Maia, T., Manu-Pereira, M., Kalfa, T. A., Pissard, et al  
2019; 94 (1): 149–61
- **Safety and Efficacy of Mitapivat in Pyruvate Kinase Deficiency.** *The New England journal of medicine*  
Grace, R. F., Rose, C. n., Layton, D. M., Galactéros, F. n., Barcellini, W. n., Morton, D. H., van Beers, E. J., Yaish, H. n., Ravindranath, Y. n., Kuo, K. H., Sheth, S. n., Kwiatkowski, J. L., Barbier, et al  
2019; 381 (10): 933–44
- **Health Related Quality of Life and Fatigue in Patients with Pyruvate Kinase Deficiency**  
Van Beers, E. J., Kuo, K. M., Morton, D., Barcellini, W., Eber, S. W., Glader, B., Yaish, H. M., Chonat, S., Kollmar, N., Despotovic, J. M., Pospisilova, D., Knoll, C. M., Kwiatkowski, et al  
AMER SOC HEMATOLOGY.2018
- **Leucine for the Treatment of Transfusion Dependence in Patients with Diamond Blackfan Anemia**  
Vlachos, A., Atsidaftos, E., Muir, E., Rogers, Z. R., Lababidi, M., Alhushki, W., Farrar, J. E., Glader, B., Gruner, B., Hartung, H., Knoll, C. M., Nalepa, G., Narla, et al  
AMER SOC HEMATOLOGY.2018
- **Pharmacological Inhibition of Nlk (Nemo-like Kinase) Rescues Erythropoietic Defects in Pre-Clinical Models of Diamond Blackfan Anemia**  
Wilkes, M. C., Chen, J., Siva, K., Veretti, G., Dever, D. P., Youn, M., Chae, H., Mercado, J. D., Saxena, M., Narla, A., Glader, B., Porteus, M., Repellin, et al  
AMER SOC HEMATOLOGY.2018
- **Pklr Intron Splicing-Associated Mutations and Alternate Diagnoses Are Common in Pyruvate Kinase Deficient Patients with Single or No Pklr Coding Mutations**  
Lezon-Geyda, K., Rose, M. J., McNaull, M. A., Knoll, C. M., Yaish, H. M., Pastore, Y. D., Fermi, E., Glader, B., Bianchi, P., Grace, R. F., Gallagher, P. G.  
AMER SOC HEMATOLOGY.2018
- **Genotype-Response Correlation in DRIVE PK, a Phase 2 Study of AG-348 in Patients with Pyruvate Kinase Deficiency**  
Kung, C., Kosinski, P. A., Mangus, H., Hua, L., Connor, G., Mobilia, M., Sullivan, K., Frye, S., Jouvin, M., Grace, R. F., Glader, B., Bowden, C.  
AMER SOC HEMATOLOGY.2018
- **The Genetic Landscape of Diamond-Blackfan Anemia.** *American journal of human genetics*  
Ulirsch, J. C., Verboon, J. M., Kazerounian, S., Guo, M. H., Yuan, D., Ludwig, L. S., Handsaker, R. E., Abdulhay, N. J., Fiorini, C., Genovese, G., Lim, E. T., Cheng, A., Cummings, et al

2018

- **Addressing the diagnostic gaps in pyruvate kinase (PK) deficiency: Consensus recommendations on the diagnosis of PK deficiency.** *American journal of hematology*  
Bianchi, P., Elisa Fermo, E., Glader, B., Kanno, H., Agarwal, A., Barcellini, W., Eber, S., Hoyer, J. D., Kuter, D. J., Maia, T. M., Del Mar Manu-Pereira, M., Kalfa, T. A., Pissard, et al  
2018
- **Hb Adana (HBA2 or HBA1: c.179G > A) and alpha thalassemia: Genotype-phenotype correlation** *PEDIATRIC BLOOD & CANCER*  
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