

## DAFTAR PUSTAKA

- Arya, V. B. et al. 2014. "HNF4A Mutation: Switch from Hyperinsulinaemic Hypoglycaemia to Maturity-Onset Diabetes of the Young, and Incretin Response." *Diabetic Medicine* 31(3): 11–15. doi: [10.1111/dme.12369](https://doi.org/10.1111/dme.12369)
- Bazi, A., Javad SR, Daryoush R, Omolbanin SA., Amin S. 2017. "Diabetes Mellitus in Thalassemia Major Patients: A Report from the Southeast of Iran". *Journal of Clinical and Diagnostic Research* 11(5):BC01-BC04. doi: [10.7860/JCDR/2017/24762.9806](https://doi.org/10.7860/JCDR/2017/24762.9806)
- Borgna-Pignatti, Caterina, and Maria Rita Gamberini. 2011. "Complications of Thalassemia Major and Their Treatment." *Expert Review of Hematology* 4(3): 353–66. doi: [10.1586/ehm.11.29](https://doi.org/10.1586/ehm.11.29).
- Cappellini MD; Cohen A; Porter J; Taher A; Viprakasit V. 2014. "Guidelines for the Management of Transfusion Dependent Thalassemia (TDT)". Thalassemia International Federation
- Chatterjee R, Bajoria R. 2009. "New Concept in natural history and management of diabetes mellitus in thalassemia major." *Haemoglobin* 33 Suppl 1 : S127-30. doi: [10.3109/09553000903347880](https://doi.org/10.3109/09553000903347880)
- Cieza-Borella C., Gonzalo DS., Isabel MP., Manuel PD., Rogelio GS. 2014. "Early Onset Type 2 Diabetes Mellitus is Associated to HNF4A t130i Polymorphism in Families of Central Spain". *Journal of Investigative Medicine* 62(8):968-974. doi: [10.1097/JIM.000000000000114](https://doi.org/10.1097/JIM.000000000000114)
- Esposito, Breno P et al. 2003. "Labile Plasma Iron in Iron Overload: Redox Activity and Susceptibility to Chelation." *Blood* 102(7): 2670–77. doi : [10.1182/blood-2003-03-0807](https://doi.org/10.1182/blood-2003-03-0807).
- Genecards Human Gene Database. 2018. "HNF4A." [www.genecards.org/cgi-bin/carddisp.pl?gene=HNF4A](http://www.genecards.org/cgi-bin/carddisp.pl?gene=HNF4A). Diakses pada 23 Desember 2018
- Genecards Human Gene Database. 2018. "PTPN1". <http://www.genecards.org/cgi-bin/carddisp.pl?gene=PTPN1>. Diakses pada 23 Desember 2018.
- Ghosh S, Watanabe RM, Valle TT, et al. 2000. "The Finland-United States investigation of non-insulin-dependent diabetes mellitus genetics (FUSION) study. I. An autosomal genome scan for genes that predispose to type 2 diabetes." *The American Journal of Human Genetic* 67:1174–1185. DOI: [10.1016/S0002-9297\(07\)62948-6](https://doi.org/10.1016/S0002-9297(07)62948-6)
- Gupta, Rana K et al. 2005. "The MODY1 Gene HNF-4  $\alpha$  Regulates Selected Genes Involved in Insulin Secretion." *The Journal of Clinical Investigation* 115(4):1006-1015. doi: [10.1172/JCI200522365](https://doi.org/10.1172/JCI200522365)

- Gurzov EN., Stanlay WJ., Brodnicki TC., Thomas HE. 2015. "Protein tyrosine phosphatase: molecular switches in metabolism and diabetes". *Trends Endocrinol Metabolism* 26(1):30-39. DOI: [10.1016/j.tem.2014.10.004](https://doi.org/10.1016/j.tem.2014.10.004)
- Ihara, Arisa *et al.* 2005. "Functional Characterization of the HNF4 $\alpha$  Isoform (HNF4 $\alpha$ 8) Expressed in Pancreatic  $\beta$ -Cells." *Biochemical and Biophysical Research Communications* 329(3): 984–90. DOI: [10.1016/j.bbrc.2005.02.072](https://doi.org/10.1016/j.bbrc.2005.02.072)
- Hall, John E. 2016. *Guyton and Hall Textbook of medical physiology*. Philadelphia: Elsevier.
- International Diabetes Federation. 2017. *IDF Diabetes Atlas 8<sup>th</sup> Edition* 2017.
- Kapoor, Ritika R *et al.* 2008. "Maturity-Onset Diabetes of the Young Due to." *Diabetes Care* 57(June): 1659–63.
- Kasper DL., Fauci AS., Hauser SL., Longo DL, *et al.* 2015. *Harrison's principles of internal medicine 19<sup>th</sup> edition*. New York: McGraw Hill Education.
- Kementerian Kesehatan RI. 2017. "Skrining Penting untuk Cegah Thalassemia". [www.depkes.go.id/article/view/17050900002/skrining-penting-untuk-cegah-thalassemia.html](http://www.depkes.go.id/article/view/17050900002/skrining-penting-untuk-cegah-thalassemia.html) diakses 28 Juni 2018
- Li MJ., Peng SS., Lu MY., Chang HH, Yang YL., *et al.* 2014. "Diabetes mellitus in patients with thalassemia major." *Pediatr Blood Cancer* 61(1):20-24. doi: [10.1002/psc.24754](https://doi.org/10.1002/psc.24754).
- Love-Gregory, Latisha, and M A Permutt. 2007. "HNF4A Genetic Variants: Role in Diabetes. [Review] [53 Refs]." *Current Opinion in Clinical Nutrition & Metabolic Care* 10(4): 397–402. DOI: [10.1097/MCO.0b013e3281e3888d](https://doi.org/10.1097/MCO.0b013e3281e3888d)
- Love-gregory, Latisha D *et al.* 2004. "A Common Polymorphism in the Upstream Promoter Region of the Hepatocyte Nuclear Factor-4 $\alpha$  Gene on and Appears to Contribute to the Evidence for Linkage in an Ashkenazi Jewish Population." *Diabetes* 53: 1134–40. [doi.org/10.2337/diabetes.53.4.1134](https://doi.org/10.2337/diabetes.53.4.1134)
- Mackawy AMH, Abd-Alfarag, Ahmed E, Badawy MEH. 2016. "Association of Protein Tyrosine Phosphatase 1B (PTPN1) Gene Polymorphisms (1023C>A and 467T>C) With Type 2 Diabetes: A Case-Control Study". *Journal of Clinical and Medical Genomics* 3(2): 135. doi:[10.4172/2472-128X.1000135](https://doi.org/10.4172/2472-128X.1000135)
- Menjivar M., Granados-Silvestre MA., Montufar-Robles I., Herera M, Tusie-Luna MT., *et al.* 2008. "High frequency of T130I mutation of HNF4A gene in Mexican patients with early onset type 2 diabetes" *Clinical Genetics* 73(2):185-7. DOI:[10.1111/j.1399-0004.2007.00928.x](https://doi.org/10.1111/j.1399-0004.2007.00928.x)
- Meshkani R, Taghikhani M, Al-Kateb H, Larijani B, Khatami S, *et al.* 2007. "Polymorphisms within the Protein Tyrosine Phosphatase 1B (PTPN1) Gene Promoter: Functional Characterization and Association with Type 2

Diabetes and Related Metabolic Traits". *Clinical Chemistry* 53: 1585-1592.  
DOI: [10.1373/clinchem.2007.088146](https://doi.org/10.1373/clinchem.2007.088146)

Modell, Bernadette, and Matthew Darlison. 2008. "Global Epidemiology of Haemoglobin Disorders and Derived Service Indicators." *Bulletin of the World Health Organization* 86(6): 480–87. doi: [10.2471/BLT.06.036673](https://doi.org/10.2471/BLT.06.036673)

Mok A, Cao H, Zinman B, Hanley AJ, Harris SB, *et al.* 2002. "A single nucleotide polymorphism in protein tyrosine phosphatase PTP-1B is associated with protection from diabetes or impaired glucose tolerance in Oji-Cree". *Journal of Clinical Endocrinology and Metabolism* 87: 724-727. DOI:[10.1210/jcem.87.2.8253](https://doi.org/10.1210/jcem.87.2.8253)

Mowla A, Karimi M, Afrasiabi A, De Sanctis V., *et al.* 2002. "Prevalence of Diabetes Mellitus and Impaired Glucose Tolerance in Beta-Thalassemia Patients with and without Hepatitis C Virus Infection." *Pediatric Endocrinology Reviews* 2(Suppl 2): 282–84.

Muncie, Herbert L, and James Campbell. 2009. "Alpha and Beta Thalassemia." *American family physician* 80(4): 339–44.

National Center of Biotechnology Information. 2018. "HNF4A." [www.ncbi.nlm.nih.gov/gene/3172](http://www.ncbi.nlm.nih.gov/gene/3172) diakses pada 23 Desember 2018

National Center of Biotechnology. 2018 "rs1800961". <https://www.ncbi.nlm.nih.gov/snp/rs1800961> diakses pada 23 Desember 2018

National Center of Biotechnology. 2018 "rs612609". <https://www.ncbi.nlm.nih.gov/snp/rs612609> diakses pada 23 Desember 2018

NCBI Reference Sequences (RefSeq). "PTPN1 protein tyrosine phosphatase, non-receptor type 1 [*Homo sapiens* (human)]". <http://www.ncbi.nlm.nih.gov/gene/5770/> diakses pada 23 Desember 2018

Odom, Duncan T *et al.* 2004. "Control of Pancreas and Liver Gene Expression by HNF Transcription Factors." *Science* 303(5662): 1378–81. doi: [10.1126/science.1089769](https://doi.org/10.1126/science.1089769)

Papakonstantinou, Olympia, *et al.* 2007. "The Pancreas in  $\beta$ -Thalassemia Major : MR Imaging Features and Correlation with Iron Stores and Glucose Disturbances." *European Radiology* 17: 1535–43. DOI [10.1007/s00330-006-0507-8](https://doi.org/10.1007/s00330-006-0507-8).

Pearson, Ewan R., *et al.* 2007. "Macrosomia and Hyperinsulinaemic Hypoglycaemia in Patients with Heterozygous Mutations in the HNF4A Gene." *PLoS Medicine* 4(4): 760–69. <https://doi.org/10.1371/journal.pmed.0040118>

- Pihoker, Catherine *et al.* 2013. "Prevalence, Characteristics and Clinical Diagnosis of Maturity Onset Diabetes of the Young due to Mutations in HNF1A, HNF4A, and Glucokinase: Results from the SEARCH for Diabetes in Youth." *Journal of Clinical Endocrinology and Metabolism* 98(10): 4055–62. doi: 10.1210/jc.2013-1279
- Riset Kesehatan Dasar. 2007. *Laporan Nasional 2007*. Jakarta: Badan Penelitian dan Pengembangan Kesehatan Departemen Kesehatan Republik Indonesia
- Rujito L, Setyono J, dan Siswandari W. 2012. *Data Penelitian Project Riset Unggulan Unsoed (RUKU) 2012*. [Laporan]. Universitas Kenderal Soedirman
- Saladin KS, Stephen JS, Christina AG. 2015. *Anatomy & physiology: the unity of form and function*. New York: McGraw-Hill Education.
- Silander, K., Mohlke, K. L., Scott, L. J., Peck, E. C., Hollstein, P., Skol, A. D. 2004. "Genetic Variation Near the Hepatocyte Nuclear Factor-4 $\alpha$  Gene Predicts Susceptibility to Type 2 Diabetes." *Diabetes* 53(4): 1141–49. <https://doi.org/10.2337/diabetes.53.4.1141>
- Spencer-Jones NJ, Wang X, Snieder H, Spector TD, Carter ND, O'Dell SD. 2005. "Protein tyrosine phosphatase-1B gene *PTPNI*: selection of tagging single nucleotide polymorphisms and association with body fat, insulin sensitivity, and the metabolic syndrome in a normal female population". *Diabetes* 54 : 3296–304. <https://doi.org/10.2337/diabetes.54.11.3296>
- Thomas H, Jaschowitz K, Bulman M, Frayling TM, Mitchell SM, Roosen S, Lingott-Frieg A, Tack CJ, Ellard S, Ryffel GU, Hattersley AT. 2001. "A Distant Upstream Promoter of the HNF-4 $\alpha$  Gene Connects the Transcription Factors Involved in Maturity-Onset Diabetes of the Young." *Human Molecular Genetics*. 10(19): 2089–97.
- Tortora, GJ and Bryan Derrickson. 2014. *Principles of Anatomy and Physiology* 14<sup>th</sup> Edition. New York: John Wiley & Sons Inc.
- Weidlich, Diana, Panos Kefalas, and Julian F. Guest. 2016. "Healthcare Costs and Outcomes of Managing  $\beta$ -Thalassemia Major over 50 Years in the United Kingdom." *Transfusion* 56(5): 1038–45. DOI:[10.1111/trf.13513](https://doi.org/10.1111/trf.13513)
- Yamagata K, Furuta H, Oda N, Kaisaki PJ, Menzel S, Cox NJ, Fajans SS., Signorini S., Stoffel M, Bell GI. 1996. "Mutations in the Hepatocyte Nuclear Factor-4 $\alpha$  Gene in Maturity-Onset Diabetes of the Young (MODY1)." *Nature* 384: 458–60. DOI:[10.1038/384458a0](https://doi.org/10.1038/384458a0)
- Yip S-C, Saha S, Chernoff J. 2010. "PTP1B: a double agent in metabolism and oncogenesis". *Trends in Biochemical Sciences* 35: 442–449. doi: 10.1016/j.tibs.2010.03.004