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DAFTAR PUSTAKA

Abu Sa'd, J., Indelman, M., Pfendner, E., Falik-Zaccai, T. C., Mizrachi-Koren, M., Shalev, S., Ben Amitai, D., *et al.* 2006. Molecular epidemiology of hereditary epidermolysis bullosa in a middle eastern population. *J Invest Dermatol,* 126**;** 777-781.

Ahmed, F. E., Jeffries, C. D., Vos, P. W., Flake, G., Nuovo, G. J., Sinar, D. R., Naziri, W., *et al.* 2009. Diagnostic microrna markers for screening sporadic human colon cancer and active ulcerative colitis in stool and tissue. *Cancer Genomics Proteomics,* 6**;** 281-295.

Alverdy, J. C., Hyoju, S. K., Weigerinck, M. dan Gilbert, J. A. 2017. The gut microbiome and the mechanism of surgical infection. *Br J Surg,* 104**;** e14-e23.

Ashton, G. H., Mellerio, J. E., Dunnill, M. G., Pulkkinen, L., Christiano, A. M., Uitto, J., Eady, R. A., *et al.* 1997. A recurrent laminin 5 mutation in british patients with lethal (herlitz) junctional epidermolysis bullosa: Evidence for a mutational hotspot rather than propagation of an ancestral allele. *Br J Dermatol,* 136**;** 674-677.

Baardman, R., Yenamandra, V. K., Duipmans, J. C., Pasmooij, A. M. G., Jonkman, M. F., van den Akker, P. C. dan Bolling, M. C. 2021. Novel insights into the epidemiology of epidermolysis bullosa (eb) from the dutch eb registry: Eb more common than previously assumed? *J Eur Acad Dermatol Venereol,* 35**;** 995-1006.

Backhed, F., Roswall, J., Peng, Y., Feng, Q., Jia, H., Kovatcheva-Datchary, P., Li, Y., *et al.* 2015. Dynamics and stabilization of the human gut microbiome during the first year of life. *Cell Host Microbe,* 17**;** 690-693.

Balato, A., Cacciapuoti, S., Di Caprio, R., Marasca, C., Masara, A., Raimondo, A. dan Fabbrocini, G. 2019. Human microbiome: Composition and role in inflammatory skin diseases. *Arch Immunol Ther Exp (Warsz),* 67**;** 1-18.

Baldovino, S., Moliner, A. M., Taruscio, D., Daina, E. dan Roccatello, D. 2016. Rare diseases in europe: From a wide to a local perspective. *Isr Med Assoc J,* 18**;** 359-363.

Bardhan, A., Bruckner-Tuderman, L., Chapple, I. L. C., Fine, J. D., Harper, N., Has, C., Magin, T. M., *et al.* 2020. Epidermolysis bullosa. *Nat Rev Dis Primers,* 6**;** 27.

Bartee, L. 2019. Types of mutations. Available: https://openoregon.pressbooks.pub/mhccbiology102/chapter/7-4-what-kinds-of-gene-mutations-are-possible/ [Accessed 29 Agustus 2019].

Bereswill, S., Ekmekciu, I., Escher, U., Fiebiger, U., Stingl, K. dan Heimesaat, M. M. 2017. Lactobacillus johnsonii ameliorates intestinal, extra-intestinal and

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GADJAH MADA
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systemic pro-inflammatory immune responses following murine campylobacter jejuni infection. *Sci Rep,* 7**;** 2138.

Bishnoi, P., Ng, Y. Z., Wei, H., Tan, E. C., Lunny, D. P., Wong, X., Kin Fon, L., *et al.* 2021. Self-improving dystrophic epidermolysis bullosa: First report of clinical, molecular, and genetic characterization of five patients from southeast asia. *Am J Med Genet A,* 185**;** 625-630.

Blanton, L. V., Charbonneau, M. R., Salih, T., Barratt, M. J., Venkatesh, S., Ilkaveya, O., Subramanian, S., *et al.* 2016. Gut bacteria that prevent growth impairments transmitted by microbiota from malnourished children. *Science,* 351**;** 1-17.

Bolling, M. C., Lemmink, H. H., Jansen, G. H. dan Jonkman, M. F. 2011. Mutations in krt5 and krt14 cause epidermolysis bullosa simplex in 75% of the patients. *Br J Dermatol,* 164**;** 637-644.

Brohem, C. A., Cardeal, L. B., Tiago, M., Soengas, M. S., Barros, S. B. dan Maria-Engler, S. S. 2011. Artificial skin in perspective: Concepts and applications. *Pigment Cell Melanoma Res,* 24**;** 35-50.

Brooke, M. A., Nitoiu, D. dan Kelsell, D. P. 2012. Cell-cell connectivity: Desmosomes and disease. *J Pathol,* 226**;** 158-171.

Bruckner-Tuderman, L., Hopfner, B. dan Hammami-Hauasli, N. 1999. Biology of anchoring fibrils: Lessons from dystrophic epidermolysis bullosa. *Matrix Biol,* 18**;** 43-54.

Bruckner-Tuderman, L. dan Payne, A. S. 2019. Epidermal and dermal adhesion. *In:* Kang, S., Amagai, M., A.L., B., A.H., E., D.J., M., A.J., M. dan J.S., O. (eds.) *Fitzpatrick's dermatology 9th edition.* New York: McGraw-Hill Education.233-248.

Buford, T. W. 2017. (dis)trust your gut: The gut microbiome in age-related inflammation, health, and disease. *Microbiome,* 5**;** 80.

Cane, L. B. 1909. Epidermolysis bullosa: Three cases: With history of the disease in four generations of the same family. *Br Med J,* 1**;** 1114-1116.

Castelino, M., Eyre, S., Moat, J., Fox, G., Martin, P., Ho, P., Upton, M., *et al.* 2017. Optimisation of methods for bacterial skin microbiome investigation: Primer selection and comparison of the 454 versus miseq platform. *BMC Microbiol,* 17**;** 23.

Chong, S. C., Hon, K. L., Yuen, L. Y. P., Choi, P. C. L., Ng, W. G. G. dan Chiu, T. W. 2021. Neonatal epidermolysis bullosa: Lessons to learn about genetic counseling. *J Dermatolog Treat,* 32**;** 29-32.

Cifuentes, L., Kiritsi, D., Chen, W., Pennino, J., Ring, J., Weidinger, S. dan Has, C. 2013. A case of junctional epidermolysis bullosa with prurigo-like lesions and reduction of collagen xvii and filaggrin. *Br J Dermatol,* 169**;** 195-198.

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GADJAH MADA
Variasi klinis, mutasi genetik, dan mikrobiom saluran cerna pada epidermolisis bulosa
SUCI WIDHIATI, Prof. Dr. Hardyanto Soebono, dr., SpKK(K)
Universitas Gadjah Mada, 2022 | Diunduh dari http://etd.repository.ugm.ac.id/

Ciubotaru, D., Bergman, R., Baty, D., Indelman, M., Pfendner, E., Petronius, D., Moualem, H., *et al.* 2003. Epidermolysis bullosa simplex in israel: Clinical and genetic features. *Arch Dermatol,* 139**;** 498-505.

Clark, A. K., Haas, K. N. dan Sivamani, R. K. 2017. Edible plants and their influence on the gut microbiome and acne. *Int J Mol Sci,* 18.

Condorelli, A. G., Logli, E., Cianfarani, F., Teson, M., Diociaiuti, A., El Hachem, M., Zambruno, G., *et al.* 2019. Microrna-145-5p regulates fibrotic features of recessive dystrophic epidermolysis bullosa skin fibroblasts. *Br J Dermatol,* 181**;** 1017-1027.

Cong, J. dan Zhang, X. 2018. How human microbiome talks to health and disease. *Eur J Clin Microbiol Infect Dis,* 37**;** 1595-1601.

Cresci, G. A. dan Bawden, E. 2015. Gut microbiome: What we do and don't know. *Nutr Clin Pract,* 30**;** 734-746.

Cross, H. E., Wells, R. S. dan Esterly, J. R. 1968. Inheritance in epidermolysis bullosa letalis. *J Med Genet,* 5**;** 189-196.

Cserhalmi-Friedman, P. B., Tang, Y., Adler, A., Krey, L., Grifo, J. A. dan Christiano, A. M. 2000. Preimplantation genetic diagnosis in two families at risk for recurrence of herlitz junctional epidermolysis bullosa. *Exp Dermatol,* 9**;** 290-297.

Cserhalmi-Friedman, P. B., Yeboa, K. A. dan Christiano, A. M. 2001. DNA based molecular analysis in the rapid diagnosis of herlitz junctional epidermolysis bullosa. *Clin Exp Dermatol,* 26**;** 205-207.

Dei-Cas, I., Giliberto, F., Luce, L., Dopazo, H. dan Penas-Steinhardt, A. 2020. Metagenomic analysis of gut microbiota in non-treated plaque psoriasis patients stratified by disease severity: Development of a new psoriasis-microbiome index. *Sci Rep,* 10**;** 1-11.

Diana, I. A. Debra indonesia, chalanges and inovation. Regional Dermatology Conferences, 2017 Surabaya. PERDOSKI.

Diana, I. A. 2021. Perdoski peringati epidermolysis bulosa awareness month. Jakarta www.klikdokter.com.

Diana, I. A., Gondokaryono, S. P., Dwiyana, R. F., Rahardja, J. H., Yogya, Y. dan Gunawan, H. 2019. Intrafamilial diversity of clinical severity of epidermolysis bullosa: Case series of three generations. *The Open Dermatology Journal***;** 5.

Durant, L., Stentz, R., Noble, A., Brooks, J., Gicheva, N., Reddi, D., O'Connor, M. J., *et al.* 2020. Bacteroides thetaiotaomicron-derived outer membrane vesicles promote regulatory dendritic cell responses in health but not in inflammatory bowel disease. *Microbiome,* 8**;** 1-16.

Endo, M., Zoltick, P. W., Radu, A., Jiang, Q., Matsui, C., Marinkovich, P. M., McGrath, J., *et al.* 2012. Early intra-amniotic gene transfer using lentiviral

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GADJAH MADA
Variasi klinis, mutasi genetik, dan mikrobiom saluran cerna pada epidermolisis bulosa
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Universitas Gadjah Mada, 2022 | Diunduh dari http://etd.repository.ugm.ac.id/

vector improves skin blistering phenotype in a murine model of herlitz junctional epidermolysis bullosa. *Gene Ther,* 19**;** 561-569.

Farokhforghani, S., Fatemi, M. J., Ghanooni, P., Asadpour, F., Araghi, S. dan Nouri, A. 2021. Epidermolysis bullosa registry data in iran. *World J Plast Surg,* 10**;** 99-103.

Fassihi, H., Wessagowit, V., Ashton, G. H., Moss, C., Ward, R., Denyer, J., Mellerio, J. E., *et al.* 2005. Complete paternal uniparental isodisomy of chromosome 1 resulting in herlitz junctional epidermolysis bullosa. *Clin Exp Dermatol,* 30**;** 71-74.

Featherstone, C. 2007. Epidermolysis bullosa: From fundamental molecular biology to clinical therapies. *J Invest Dermatol,* 127**;** 256-259.

Featherstone, C. dan Uitto, J. 2007. Ex vivo gene therapy cures a blistering skin disease. *Trends Mol Med,* 13**;** 219-222.

Fine, J. D. 2016. Epidemiology of inherited epidermolysis bullosa based on incidence and prevalence estimates from the national epidermolysis bullosa registry. *JAMA Dermatol,* 152**;** 1231-1238.

Fine, J. D., Bruckner-Tuderman, L., Eady, R. A., Bauer, E. A., Bauer, J. W., Has, C., Heagerty, A., *et al.* 2014. Inherited epidermolysis bullosa: Updated recommendations on diagnosis and classification. *J Am Acad Dermatol,* 70**;** 1103-1126.

Fine, J. D. dan Mellerio, J. E. 2009a. Extracutaneous manifestations and complications of inherited epidermolysis bullosa: Part i. Epithelial associated tissues. *J Am Acad Dermatol,* 61**;** 367-384; quiz 385-366.

Fine, J. D. dan Mellerio, J. E. 2009b. Extracutaneous manifestations and complications of inherited epidermolysis bullosa: Part ii. Other organs. *J Am Acad Dermatol,* 61**;** 387-402; quiz 403-384.

Fong, K., Bailey, C. V., Tuttle, P., Cunningham, B., McGrath, J. A. dan Cho, R. J. 2017. Questioning the clinical utility of exome sequencing in developing countries. *Pediatr Dermatol,* 34**;** e32-e34.

Fozia, F., Nazli, R., Bibi, N., Khan, S. A., Muhammad, N., Shakeeb, N., Khan, S., *et al.* 2021. Whole exome sequencing confirms molecular diagnostics of three pakhtun families with autosomal recessive epidermolysis bullosa. *Front Pediatr,* 9**;** 727288.

Freeman, E. B., Köglmeier, J., Martinez, A. E., Mellerio, J. E., Haynes, L., Sebire, N. J., Lindley, K. J., *et al.* 2008. Gastrointestinal complications of epidermolysis bullosa in children. *Br J Dermatol,* 158**;** 1308-1314.

Fuentes, I., Guttmann-Gruber, C., Tay, A. S. L., Pinon Hofbauer, J., Denil, S., Reichelt, J., Palisson, F., *et al.* 2018. Reduced microbial diversity is a feature of recessive dystrophic epidermolysis bullosa-involved skin and wounds. *J Invest Dermatol,* 138**;** 2492-2495.

UNIVERSITAS
GADJAH MADA
Variasi klinis, mutasi genetik, dan mikrobiom saluran cerna pada epidermolisis bulosa
SUCI WIDHIATI, Prof. Dr. Hardyanto Soebono, dr., SpKK(K)
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Gache, Y., Allegra, M., Bodemer, C., Pisani-Spadafora, A., de Prost, Y., Ortonne, J. P. dan Meneguzzi, G. 2001. Genetic bases of severe junctional epidermolysis bullosa presenting spontaneous amelioration with aging. *Hum Mol Genet,* 10**;** 2453-2461.

Galehdari, H., Mohammadian, G., Azmoon, S., Salehi, B. dan Pedram, M. 2010. A novel col7a1 gene mutation in an iranian individual suffering dystrophic epidermolysis bullosa. *J Mol Diagn,* 12**;** 377-379.

Gao, Z., Perez-Perez, G. I., Chen, Y. dan Blaser, M. J. 2010. Quantitation of major human cutaneous bacterial and fungal populations. *J Clin Microbiol,* 48**;** 3575-3581.

Garza, L. 2019. Developmental biology of the skin. *In:* Kang, S., Amagai, M., A.L., B., A.H., E., D.J., M., A.J., M. dan J.S., O. (eds.) *Fitzpatrick's dermatology.* 9th ed. New York: McGraw-Hill Education.53-55.

Gedde-Dahl, T. J. 1971. *Epidermolysis bullosa. A clinical, genetic and epidemiologic study.,* Baltimore, Johns Hopkins University Press.

Grice, E. A. 2014. The skin microbiome: Potential for novel diagnostic and therapeutic approaches to cutaneous disease. *Semin Cutan Med Surg,* 33**;** 98-103.

Grice, E. A., Kong, H. H., Conlan, S., Deming, C. B., Davis, J., Young, A. C., Program, N. C. S., *et al.* 2009. Topographical and temporal diversity of the human skin microbiome. *Science,* 324**;** 1190-1192.

Halim-Fikri, H., Etemad, A., Abdul Latif, A. Z., Merican, A. F., Baig, A. A., Annuar, A. A., Ismail, E., *et al.* 2015. The first malay database toward the ethnic-specific target molecular variation. *BMC Res Notes,* 8**;** 176.

Hamady, M. dan Knight, R. 2009. Microbial community profiling for human microbiome projects: Tools, techniques, and challenges. *Genome Res,* 19**;** 1141-1152.

Hamady, M., Walker, J. J., Harris, J. K., Gold, N. J. dan Knight, R. 2008. Error-correcting barcoded primers for pyrosequencing hundreds of samples in multiplex. *Nat Methods,* 5**;** 235-237.

Hamilton, A., Tetreault, M., Dyment, D. A., Zou, R., Kernohan, K., Geraghty, M. T., Consortium, F. C., *et al.* 2016. Concordance between whole-exome sequencing and clinical sanger sequencing: Implications for patient care. *Mol Genet Genomic Med,* 4**;** 504-512.

Hammersen, J., Has, C., Naumann-Bartsch, N., Stachel, D., Kiritsi, D., Soder, S., Tardieu, M., *et al.* 2016. Genotype, clinical course, and therapeutic decision making in 76 infants with severe generalized junctional epidermolysis bullosa. *J Invest Dermatol,* 136**;** 2150-2157.

Has, C. 2011. Molecular genetic assays for inherited epidermolysis bullosa. *Clin Dermatol,* 29**;** 420-426.

UNIVERSITAS
GADJAH MADA
Variasi klinis, mutasi genetik, dan mikrobiom saluran cerna pada epidermolisis bulosa
SUCI WIDHIATI, Prof. Dr. Hardyanto Soebono, dr., SpKK(K)
Universitas Gadjah Mada, 2022 | Diunduh dari http://etd.repository.ugm.ac.id/

Has, C., Bauer, J. W., Bodemer, C., Bolling, M. C., Bruckner-Tuderman, L., Diem, A., Fine, J. D., *et al.* 2020. Consensus reclassification of inherited epidermolysis bullosa and other disorders with skin fragility. *Br J Dermatol,* 183**;** 614-627.

Has, C., Castiglia, D., del Rio, M., Diez, M. G., Piccinni, E., Kiritsi, D., Kohlhase, J., *et al.* 2011. Kindler syndrome: Extension of fermt1 mutational spectrum and natural history. *Hum Mutat,* 32**;** 1204-1212.

Has, C. dan Fischer, J. 2018. Inherited epidermolysis bullosa: New diagnostics and new clinical phenotypes. *Exp Dermatol*.

Has, C. dan Fischer, J. 2019. Inherited epidermolysis bullosa: New diagnostics and new clinical phenotypes. *Exp Dermatol,* 28**;** 1146-1152.

Has, C. dan He, Y. 2016. Research techniques made simple: Immunofluorescence antigen mapping in epidermolysis bullosa. *J Invest Dermatol,* 136**;** e65-e71.

Hee, S. W., Willis, A., Tudur Smith, C., Day, S., Miller, F., Madan, J., Posch, M., *et al.* 2017. Does the low prevalence affect the sample size of interventional clinical trials of rare diseases? An analysis of data from the aggregate analysis of clinicaltrials.Gov. *Orphanet J Rare Dis,* 12**;** 44.

Hendriks, A. C. A., Reubsaet, F. A. G., Kooistra-Smid, A., Rossen, J. W. A., Dutilh, B. E., Zomer, A. L., van den Beld, M. J. C., *et al.* 2020. Genome-wide association studies of shigella spp. And enteroinvasive escherichia coli isolates demonstrate an absence of genetic markers for prediction of disease severity. *BMC Genomics,* 21**;** 138.

Horiguchi, Y., Sawamura, D., Mori, R., Nakamura, H., Takahashi, K. dan Shimizu, H. 2005. Clinical heterogeneity of 1649delg mutation in the tail domain of keratin 5: A japanese family with epidermolysis bullosa simplex with mottled pigmentation. *J Invest Dermatol,* 125**;** 83-85.

Horn, H. M. dan Tidman, M. J. 2002. The clinical spectrum of dystrophic epidermolysis bullosa. *Br J Dermatol,* 146**;** 267-274.

Huttenhower, C., Gevers, D., Knight, R., Abubucker, S., Badger, J. H., Chinwalla, A. T. dan Creasy, H. H. 2012. Structure, function and diversity of the healthy human microbiome. *Nature,* 486**;** 207-214.

Institute, N. H. G. R. 2022. *Mutation* [Online]. Available: https://www.genome.gov/genetics-glossary/Mutation [Accessed 20 Maret 2022 2022].

James, W. D., Elston, D. M. dan Berger, T. G. 2011. Skin: Basic structure and function. *Andrews' disease of the skin clinical dermatology.* eleventh ed. United Kingdom: Elsevier.1-2.

Johnson, A. J., Vangay, P., Al-Ghalith, G. A., Hillmann, B. M., Ward, T. L., Shields-Cutler, R. R., Kim, A. D., *et al.* 2019. Daily sampling reveals personalized diet-microbiome associations in humans. *Cell Host Microbe,* 25**;** 789-802 e785.

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GADJAH MADA
Variasi klinis, mutasi genetik, dan mikrobiom saluran cerna pada epidermolisis bulosa
SUCI WIDHIATI, Prof. Dr. Hardyanto Soebono, dr., SpKK(K)
Universitas Gadjah Mada, 2022 | Diunduh dari http://etd.repository.ugm.ac.id/

Johnson, C. C. dan Ownby, D. R. 2017. The infant gut bacterial microbiota and risk of pediatric asthma and allergic diseases. *Transl Res,* 179**;** 60-70.

Kalliomaki, M., Kirjavainen, P., Eerola, E., Kero, P., Salminen, S. dan Isolauri, E. 2001. Distinct patterns of neonatal gut microflora in infants in whom atopy was and was not developing. *J Allergy Clin Immunol,* 107**;** 129-134.

Kang, T. W., Lee, J. S., Kim, S. E., Oh, S. W. dan Kim, S. C. 2010. Novel and recurrent mutations in keratin 5 and 14 in korean patients with epidermolysis bullosa simplex. *J Dermatol Sci,* 57**;** 90-94.

Khan, F. F., Khan, N., Rehman, S., Ejaz, A., Ali, U., Erfan, M., Ahmed, Z. M., *et al.* 2021. Identification and computational analysis of novel pathogenic variants in pakistani families with diverse epidermolysis bullosa phenotypes. *Biomolecules,* 11.

Kim, B. S., Jeon, Y. S. dan Chun, J. 2013. Current status and future promise of the human microbiome. *Pediatr Gastroenterol Hepatol Nutr,* 16**;** 71-79.

Kim, C. H. 2013. Host and microbial factors in regulation of t cells in the intestine. *Front Immunol,* 4**;** 141.

Kim, E. N., Harris, A. G., Bingham, L. J., Yan, W., Su, J. C. dan Murrell, D. F. 2017. A review of 52 pedigrees with epidermolysis bullosa simplex identifying ten novel mutations in krt5 and krt14 in australia. *Acta Derm Venereol,* 97**;** 1114-1119.

Kiritsi, D., Garcia, M., Brander, R., Has, C., Meijer, R., Jose Escamez, M., Kohlhase, J., *et al.* 2014. Mechanisms of natural gene therapy in dystrophic epidermolysis bullosa. *J Invest Dermatol,* 134**;** 2097-2104.

Kiritsi, D. dan Nystrom, A. 2018. Recent advances in understanding and managing epidermolysis bullosa. *F1000Res,* 7.

Kivirikko, S., McGrath, J. A., Pulkkinen, L., Uitto, J. dan Christiano, A. M. 1996. Mutational hotspots in the lamb3 gene in the lethal (herlitz) type of junctional epidermolysis bullosa. *Hum Mol Genet,* 5**;** 231-237.

Komorowski, L., Muller, R., Vorobyev, A., Probst, C., Recke, A., Jonkman, M. F., Hashimoto, T., *et al.* 2013. Sensitive and specific assays for routine serological diagnosis of epidermolysis bullosa acquisita. *J Am Acad Dermatol,* 68**;** e89-95.

Kong, H. H. 2011. Skin microbiome: Genomics-based insights into the diversity and role of skin microbes. *Trends Mol Med,* 17**;** 320-328.

Kong, H. H. dan Segre, J. A. 2017. The molecular revolution in cutaneous biology: Investigating the skin microbiome. *J Invest Dermatol,* 137**;** e119-e122.

Korge, B. P. dan Krieg, T. 1996. The molecular basis for inherited bullous diseases. *J Mol Med (Berl),* 74**;** 59-70.

UNIVERSITAS
GADJAH MADA
Variasi klinis, mutasi genetik, dan mikrobiom saluran cerna pada epidermolisis bulosa
SUCI WIDHIATI, Prof. Dr. Hardyanto Soebono, dr., SpKK(K)
Universitas Gadjah Mada, 2022 | Diunduh dari http://etd.repository.ugm.ac.id/

Kosiewicz, M. M., Dryden, G. W., Chhabra, A. dan Alard, P. 2014. Relationship between gut microbiota and development of t cell associated disease. *FEBS Lett,* 588**;** 4195-4206.

Kragsnaes, M. S., Kjeldsen, J., Horn, H. C., Munk, H. L., Pedersen, F. M., Holt, H. M., Pedersen, J. K., *et al.* 2018. Efficacy and safety of faecal microbiota transplantation in patients with psoriatic arthritis: Protocol for a 6-month, double-blind, randomised, placebo-controlled trial. *BMJ Open,* 8**;** e019231.

Lai-Cheong, J. E., Parsons, M., Tanaka, A., Ussar, S., South, A. P., Gomathy, S., Mee, J. B., *et al.* 2009. Loss-of-function fermt1 mutations in kindler syndrome implicate a role for fermitin family homolog-1 in integrin activation. *Am J Pathol,* 175**;** 1431-1441.

Lechler, T. 2019. Growth and differentiation of the epidermis. *In:* Kang, S., Amagai, M., A.L., B., A.H., E., D.J., M., A.J., M. dan J.S., O. (eds.) *Fitzpatrick's dermatology 9th edition.* 9th ed. New York: McGraw-Hill Education.62-69.

Lin, Z., Li, S., Feng, C., Yang, S., Wang, H., Ma, D., Zhang, J., *et al.* 2016. Stabilizing mutations of klhl24 ubiquitin ligase cause loss of keratin 14 and human skin fragility. *Nat Genet,* 48**;** 1508-1516.

Lincoln, V., Cogan, J., Hou, Y., Hirsch, M., Hao, M., Alexeev, V., De Luca, M., *et al.* 2018. Gentamicin induces lamb3 nonsense mutation readthrough and restores functional laminin 332 in junctional epidermolysis bullosa. *Proc Natl Acad Sci U S A,* 115**;** E6536-e6545.

Lu, S., Wang, J., Chitsaz, F., Derbyshire, M. K., Geer, R. C., Gonzales, N. R., Gwadz, M., *et al.* 2020. Cdd/sparcle: The conserved domain database in 2020. *Nucleic Acids Res,* 48**;** d265-d268.

Lu, T., Chen, Y., Guo, Y., Sun, J., Shen, W., Yuan, M., Zhang, S., *et al.* 2019. Altered gut microbiota diversity and composition in chronic urticaria. *Dis Markers,* 2019**;** 6417471.

Lucky, A. W., Dagaonkar, N., Lammers, K., Husami, A., Kissell, D. dan Zhang, K. 2018. A comprehensive next-generation sequencing assay for the diagnosis of epidermolysis bullosa. *Pediatr Dermatol,* 35**;** 188-197.

Mahajan, R., Manjunath, S., Madakshira, M. G., Chatterjee, D., Bishnoi, A., De, D., Handa, S., *et al.* 2022. Correlating clinical and laboratory diagnostic modalities in the diagnosis of epidermolysis bullosa in a resource-poor setting. *J Cutan Pathol,* 49**;** 454-459.

Mallipeddi, R., Bleck, O., Mellerio, J. E., Ashton, G. H., Eady, R. A. dan McGrath, J. A. 2003. Dilemmas in distinguishing between dominant and recessive forms of dystrophic epidermolysis bullosa. *Br J Dermatol,* 149**;** 810-818.

Margolis, R. J. 2016. Utilizing innovative statistical methods and trial design in rare disease settings. Duke Center for Health Policy.

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GADJAH MADA
Variasi klinis, mutasi genetik, dan mikrobiom saluran cerna pada epidermolisis bulosa
SUCI WIDHIATI, Prof. Dr. Hardyanto Soebono, dr., SpKK(K)
Universitas Gadjah Mada, 2022 | Diunduh dari http://etd.repository.ugm.ac.id/

Mariath, L. M., Santin, J. T., Frantz, J. A., Doriqui, M. J. R., Kiszewski, A. E. dan Schuler-Faccini, L. 2019. An overview of the genetic basis of epidermolysis bullosa in brazil: Discovery of novel and recurrent disease-causing variants. *Clin Genet,* 96**;** 189-198.

Mariath, L. M., Santin, J. T., Schuler-Faccini, L. dan Kiszewski, A. E. 2020. Inherited epidermolysis bullosa: Update on the clinical and genetic aspects. *An Bras Dermatol,* 95**;** 551-569.

Martin, A. R., Williams, E., Foulger, R. E., Leigh, S., Daugherty, L. C., Niblock, O., Leong, I. U. S., *et al.* 2019. Panelapp crowdsources expert knowledge to establish consensus diagnostic gene panels. *Nat Genet,* 51**;** 1560-1565.

McGrath, J. A. 2017. A new clinical diagnostic matrix for epidermolysis bullosa. *Br J Dermatol,* 176**;** 1442-1443.

McMillan, J. R., McGrath, J. A., Tidman, M. J. dan Eady, R. A. 1998. Hemidesmosomes show abnormal association with the keratin filament network in junctional forms of epidermolysis bullosa. *J Invest Dermatol,* 110**;** 132-137.

Mellerio, J. E., Dunnill, M. G., Allison, W., Ashton, G. H., Christiano, A. M., Uitto, J., Eady, R. A., *et al.* 1997. Recurrent mutations in the type vii collagen gene (col7a1) in patients with recessive dystrophic epidermolysis bullosa. *J Invest Dermatol,* 109**;** 246-249.

Meyer-Mueller, C., Osborn, M. J., Tolar, J., Boull, C. dan Ebens, C. L. 2022. Revertant mosaicism in epidermolysis bullosa. *Biomedicines,* 10.

Minakawa, S., Nakano, H., Nakajima, K., Matsuzaki, Y., Takiyoshi, N., Akasaka, E., Rokunohe, D., *et al.* 2013. Mutational analysis on 16 japanese population cases with epidermolysis bullosa simplex. *J Dermatol Sci,* 72**;** 330-332.

Morales, P., Fujio, S., Navarrete, P., Ugalde, J. A., Magne, F., Carrasco-Pozo, C., Tralma, K., *et al.* 2016. Impact of dietary lipids on colonic function and microbiota: An experimental approach involving orlistat-induced fat malabsorption in human volunteers. *Clin Transl Gastroenterol,* 7**;** e161.

Murata, T., Masunaga, T., Ishiko, A., Shimizu, H. dan Nishikawa, T. 2004. Differences in recurrent col7a1 mutations in dystrophic epidermolysis bullosa: Ethnic-specific and worldwide recurrent mutations. *Arch Dermatol Res,* 295**;** 442-447.

Nagao-Watanabe, M., Fukao, T., Matsui, E., Kaneko, H., Inoue, R., Kawamoto, N., Kasahara, K., *et al.* 2004. Identification of somatic and germline mosaicism for a keratin 5 mutation in epidermolysis bullosa simplex in a family of which the proband was previously regarded as a sporadic case. *Clin Genet,* 66**;** 236-238.

Nagy, N. dan McGrath, J. A. 2010. Blistering skin diseases: A bridge between dermatopathology and molecular biology. *Histopathology,* 56**;** 91-99.

UNIVERSITAS
GADJAH MADA
Variasi klinis, mutasi genetik, dan mikrobiom saluran cerna pada epidermolisis bulosa
SUCI WIDHIATI, Prof. Dr. Hardyanto Soebono, dr., SpKK(K)
Universitas Gadjah Mada, 2022 | Diunduh dari http://etd.repository.ugm.ac.id/

Nakano, A., Lestringant, G. G., Paperna, T., Bergman, R., Gershoni, R., Frossard, P., Kanaan, M., *et al.* 2002. Junctional epidermolysis bullosa in the middle east: Clinical and genetic studies in a series of consanguineous families. *J Am Acad Dermatol,* 46**;** 510-516.

Natsuga, K., Nishie, W., Shinkuma, S., Nakamura, H., Matsushima, Y., Tatsuta, A., Komine, M., *et al.* 2011a. Expression of exon-8-skipped kindlin-1 does not compensate for defects of kindler syndrome. *J Dermatol Sci,* 61**;** 38-44.

Natsuga, K., Nishie, W., Smith, B. J., Shinkuma, S., Smith, T. A., Parry, D. A., Oiso, N., *et al.* 2011b. Consequences of two different amino-acid substitutions at the same codon in krt14 indicate definitive roles of structural distortion in epidermolysis bullosa simplex pathogenesis. *J Invest Dermatol,* 131**;** 1869-1876.

Naussbaum, R. L., McInnes, R. R., Willard, H. F., Hamosh, A. dan Thompson, M. W. 2007. *Thompson & thompson genetics in medicine,* Philadelphia, Saunders/Elsevier.

Niguidula, N., Alamillo, C., Shahmirzadi Mowlavi, L., Powis, Z., Cohen, J. S. dan Farwell Hagman, K. D. 2018. Clinical whole-exome sequencing results impact medical management. *Mol Genet Genomic Med,* 6**;** 1068-1078.

O'Neill, C. A., Monteleone, G., McLaughlin, J. T. dan Paus, R. 2016. The gut-skin axis in health and disease: A paradigm with therapeutic implications. *Bioessays,* 38**;** 1167-1176.

Oliveira, Z. N., Perigo, A. M., Fukumori, L. M. dan Aoki, V. 2010. Immunological mapping in hereditary epidermolysis bullosa. *An Bras Dermatol,* 85**;** 856-861.

Pan, F., Zhang, L., Li, M., Hu, Y., Zeng, B., Yuan, H., Zhao, L., *et al.* 2018. Predominant gut lactobacillus murinus strain mediates anti-inflammaging effects in calorie-restricted mice. *Microbiome,* 6**;** 1-17.

Pasmooij, A. M., Garcia, M., Escamez, M. J., Nijenhuis, A. M., Azon, A., Cuadrado-Corrales, N., Jonkman, M. F., *et al.* 2010. Revertant mosaicism due to a second-site mutation in col7a1 in a patient with recessive dystrophic epidermolysis bullosa. *J Invest Dermatol,* 130**;** 2407-2411.

Pasmooij, A. M., Pas, H. H., Deviaene, F. C., Nijenhuis, M. dan Jonkman, M. F. 2005. Multiple correcting col17a1 mutations in patients with revertant mosaicism of epidermolysis bullosa. *Am J Hum Genet,* 77**;** 727-740.

Patel, T., Bhattacharya, P. dan Das, S. 2016. Gut microbiota: An indicator to gastrointestinal tract diseases. *J Gastrointest Cancer,* 47**;** 232-238.

Pearson, R. W. 1962. Studies on the pathogenesis of epidermolysis bullosa. *J Invest Dermatol,* 39**;** 551-575.

Pérez-Cobas, A. E., Gomez-Valero, L. dan Buchrieser, C. 2020. Metagenomic approaches in microbial ecology: An update on whole-genome and marker gene sequencing analyses. *Microb Genom,* 6.

UNIVERSITAS
GADJAH MADA
Variasi klinis, mutasi genetik, dan mikrobiom saluran cerna pada epidermolisis bulosa
SUCI WIDHIATI, Prof. Dr. Hardyanto Soebono, dr., SpKK(K)
Universitas Gadjah Mada, 2022 | Diunduh dari http://etd.repository.ugm.ac.id/

Pulkkinen, L., Meneguzzi, G., McGrath, J. A., Xu, Y., Blanchet-Bardon, C., Ortonne, J. P., Christiano, A. M., *et al.* 1997. Predominance of the recurrent mutation r635x in the lamb3 gene in european patients with herlitz junctional epidermolysis bullosa has implications for mutation detection strategy. *J Invest Dermatol,* 109**;** 232-237.

Rahayu, E. S., Utami, T., Mariyatun, M., Hasan, P. N., Kamil, R. Z., Setyawan, R. H., Pamungkaningtyas, F. H., *et al.* 2019. Gut microbiota profile in healthy indonesians. *World J Gastroenterol,* 25**;** 1478-1491.

Rayinda, T., van Steensel, M. dan Danarti, R. 2021. Inherited skin disorders presenting with poikiloderma. *Int J Dermatol,* 60**;** 1343-1353.

Riahi, K., Ghanbari Mardasi, F., Talebi, F., Jasemi, F. dan Mohammadi Asl, J. 2021. Digenic mutations in junctional epidermolysis bullosa in an iranian family. *Cell J,* 23**;** 598-602.

Rinninella, E., Raoul, P., Cintoni, M., Franceschi, F., Miggiano, G. A. D., Gasbarrini, A. dan Mele, M. C. 2019. What is the healthy gut microbiota composition? A changing ecosystem across age, environment, diet, and diseases. *Microorganisms,* 7.

Rosenberg, E. dan Zilber-Rosenberg, I. 2019. The hologenome concept of evolution: Medical implications. *Rambam Maimonides Med J,* 10.

Salem, I., Ramser, A., Isham, N. dan Ghannoum, M. A. 2018. The gut microbiome as a major regulator of the gut-skin axis. *Front Microbiol,* 9**;** 1459.

Scher, J. U. dan Abramson, S. B. 2011. The microbiome and rheumatoid arthritis. *Nat Rev Rheumatol,* 7**;** 569-578.

Schlaeppi, K. dan Bulgarelli, D. 2015. The plant microbiome at work. *Mol Plant Microbe Interact,* 28**;** 212-217.

Schommer, N. N. dan Gallo, R. L. 2013. Structure and function of the human skin microbiome. *Trends Microbiol,* 21**;** 660-668.

Segata, N., Haake, S. K., Mannon, P., Lemon, K. P., Waldron, L., Gevers, D., Huttenhower, C., *et al.* 2012. Composition of the adult digestive tract bacterial microbiome based on seven mouth surfaces, tonsils, throat and stool samples. *Genome Biol,* 13**;** R42.

Selvanderan, S. P., Goldblatt, F., Nguyen, N. Q. dan Costello, S. P. 2019. Faecal microbiota transplantation for clostridium difficile infection resulting in a decrease in psoriatic arthritis disease activity. *Clin Exp Rheumatol,* 37**;** 514-515.

Shimizu, H., Takizawa, Y., McGrath, J. A., Pulkkinen, L., Christiano, A. M., Uitto, J., Burgeson, R. E., *et al.* 1997. Absence of r42x and r635x mutations in the lamb3 gene in 12 japanese patients with junctional epidermolysis bullosa. *Arch Dermatol Res,* 289**;** 174-176.

UNIVERSITAS
GADJAH MADA
Variasi klinis, mutasi genetik, dan mikrobiom saluran cerna pada epidermolisis bulosa
SUCI WIDHIATI, Prof. Dr. Hardyanto Soebono, dr., SpKK(K)
Universitas Gadjah Mada, 2022 | Diunduh dari http://etd.repository.ugm.ac.id/

Sims, G. E. dan Kim, S. H. 2011. Whole-genome phylogeny of escherichia coli/shigella group by feature frequency profiles (ffps). *Proc Natl Acad Sci U S A,* 108**;** 8329-8334.

Singer, H. M., Levin, L. E., Garzon, M. C., Lauren, C. T., Planet, P. J., Kittler, N. W., Whittier, S., *et al.* 2018. Wound culture isolated antibiograms and caregiver-reported skin care practices in children with epidermolysis bullosa. *Pediatr Dermatol,* 35**;** 92-96.

Song, H., Yoo, Y., Hwang, J., Na, Y. C. dan Kim, H. S. 2016. Faecalibacterium prausnitzii subspecies-level dysbiosis in the human gut microbiome underlying atopic dermatitis. *J Allergy Clin Immunol,* 137**;** 852-860.

Stawczyk-Macieja, M., Wertheim-Tysarowska, K., Jakubowski, R., Szczerkowska-Dobosz, A., Krygier, M., Wilkowska, A., Sawicka, J., *et al.* 2018. A novel de novo mutation p.Ala428asp in krt5 gene as a cause of localized epidermolysis bullosa simplex. *Exp Dermatol,* 28**;** 1131-1134.

Takeichi, T., Liu, L., Fong, K., Ozoemena, L., McMillan, J. R., Salam, A., Campbell, P., *et al.* 2015a. Whole-exome sequencing improves mutation detection in a diagnostic epidermolysis bullosa laboratory. *Br J Dermatol,* 172**;** 94-100.

Takeichi, T., Nanda, A., Liu, L., Aristodemou, S., McMillan, J. R., Sugiura, K., Akiyama, M., *et al.* 2015b. Founder mutation in dystonin-e underlying autosomal recessive epidermolysis bullosa simplex in kuwait. *Br J Dermatol,* 172**;** 527-531.

Tang, H. Y., Du, W. D., Cui, Y., Fan, X., Quan, C., Fang, Q. Y., Zhou, F. S., *et al.* 2009. One novel and two recurrent mutations in the keratin 5 gene identified in chinese patients with epidermolysis bullosa simplex. *Clin Exp Dermatol,* 34**;** e957-961.

Tenedini, E., Artuso, L., Bernardis, I., Artusi, V., Percesepe, A., De Rosa, L., Contin, R., *et al.* 2015. Amplicon-based next-generation sequencing: An effective approach for the molecular diagnosis of epidermolysis bullosa. *Br J Dermatol,* 173**;** 731-738.

Tett, A., Pasolli, E., Farina, S., Truong, D. T., Asnicar, F., Zolfo, M., Beghini, F., *et al.* 2017. Unexplored diversity and strain-level structure of the skin microbiome associated with psoriasis. *NPJ Biofilms Microbiomes,* 3**;** 14.

Thusberg, J. dan Vihinen, M. 2009. Pathogenic or not? And if so, then how? Studying the effects of missense mutations using bioinformatics methods. *Hum Mutat,* 30**;** 703-714.

Travis, S. P., McGrath, J. A., Turnbull, A. J., Schofield, O. M., Chan, O., O'Connor, A. F., Mayou, B., *et al.* 1992. Oral and gastrointestinal manifestations of epidermolysis bullosa. *Lancet,* 340**;** 1505-1506.

UNIVERSITAS
GADJAH MADA
Variasi klinis, mutasi genetik, dan mikrobiom saluran cerna pada epidermolisis bulosa
SUCI WIDHIATI, Prof. Dr. Hardyanto Soebono, dr., SpKK(K)
Universitas Gadjah Mada, 2022 | Diunduh dari http://etd.repository.ugm.ac.id/

Turnbaugh, P. J., Ley, R. E., Mahowald, M. A., Magrini, V., Mardis, E. R. dan Gordon, J. I. 2006. An obesity-associated gut microbiome with increased capacity for energy harvest. *Nature,* 444**;** 1027-1031.

Vahidnezhad, H., Youssefian, L., Saeidian, A. H., Mozafari, N., Barzegar, M., Sotoudeh, S., Daneshpazhooh, M., *et al.* 2016. Krt5 and krt14 mutations in epidermolysis bullosa simplex with phenotypic heterogeneity, and evidence of semidominant inheritance in a multiplex family. *J Invest Dermatol,* 136**;** 1897-1901.

Vahidnezhad, H., Youssefian, L., Saeidian, A. H., Touati, A., Pajouhanfar, S., Baghdadi, T., Shadmehri, A. A., *et al.* 2019a. Mutations in plod3, encoding lysyl hydroxylase 3, cause a complex connective tissue disorder including recessive dystrophic epidermolysis bullosa-like blistering phenotype with abnormal anchoring fibrils and type vii collagen deficiency. *Matrix Biol,* 81**;** 91-106.

Vahidnezhad, H., Youssefian, L., Saeidian, A. H. dan Uitto, J. 2019b. Phenotypic spectrum of epidermolysis bullosa: The paradigm of syndromic versus non-syndromic skin fragility disorders. *J Invest Dermatol,* 139**;** 522-527.

Vanden Oever, M., Muldoon, D., Mathews, W., McElmurry, R. dan Tolar, J. 2016. Mir-29 regulates type vii collagen in recessive dystrophic epidermolysis bullosa. *J Invest Dermatol,* 136**;** 2013-2021.

Varki, R., Sadowski, S., Pfendner, E. dan Uitto, J. 2006. Epidermolysis bullosa. I. Molecular genetics of the junctional and hemidesmosomal variants. *J Med Genet,* 43**;** 641-652.

Viennois, E., Chassaing, B., Tahsin, A., Pujada, A., Wang, L., Gewirtz, A. T. dan Merlin, D. 2019. Host-derived fecal microrna*s* can indicate gut microbiota healthiness and ability to induce inflammation. *Theranostics,* 9**;** 4542-4557.

von Bartenwerffer, W., Has, C., Arin, M. J., Tantcheva-Poor, I., Kreuter, A., Kremer, K., Arshah, T., *et al.* 2011. Mild recessive dystrophic epidermolysis bullosa associated with two compound heterozygous col7a1 mutations. *Eur J Dermatol,* 21**;** 170-172.

Wagner, J. E., Ishida-Yamamoto, A., McGrath, J. A., Hordinsky, M., Keene, D. R., Woodley, D. T., Chen, M., *et al.* 2010. Bone marrow transplantation for recessive dystrophic epidermolysis bullosa. *N Engl J Med,* 363**;** 629-639.

Wang, H., Yang, Y., Zhou, J., Cao, J., He, X., Li, L., Gao, S., *et al.* 2018. Targeted next-generation sequencing identifies a novel mutation of lamb3 in a chinese neonatal patient presented with junctional epidermolysis bullosa. *Medicine (Baltimore),* 97**;** e13225.

Watt, S. A., Dayal, J. H., Wright, S., Riddle, M., Pourreyron, C., McMillan, J. R., Kimble, R. M., *et al.* 2015. Lysyl hydroxylase 3 localizes to epidermal basement membrane and is reduced in patients with recessive dystrophic epidermolysis bullosa. *PLoS One,* 10**;** e0137639.

UNIVERSITAS
GADJAH MADA
Variasi klinis, mutasi genetik, dan mikrobiom saluran cerna pada epidermolisis bulosa
SUCI WIDHIATI, Prof. Dr. Hardyanto Soebono, dr., SpKK(K)
Universitas Gadjah Mada, 2022 | Diunduh dari http://etd.repository.ugm.ac.id/

Webber, B. R. dan Tolar, J. 2015. From marrow to matrix: Novel gene and cell therapies for epidermolysis bullosa. *Mol Ther,* 23**;** 987-992.

Widhiati, S., Danarti, R., Trisnowati, N., Purnomosari, D., Wibawa, T. dan Soebono, H. 2021a. Novel mutations of epidermolysis bullosa identified using whole-exome sequencing in indonesian javanese patients. *Intractable Rare Dis Res,* 10**;** 88-94.

Widhiati, S., Marcella, B., Dewi, S., Paramitasari, A. R., Ellistari, E. Y. dan Julianto, I. 2019. Clinical diagnostic matrix (cdm) as a tool to diagnose subtypes of epidermolysis bullosa cases in children. *J Gen Proced Dermatol Venereol Indones,* 3**;** 1-7.

Widhiati, S., Purnomosari, D., Wibawa, T. dan Soebono, H. 2021b. Gut-skin axis microbiome in various skin disorders: A literature review. *Dermatology Reports,* Ahead of print**;** 26.

Yan, Y., Meng, Z., Hao, S., Wang, F., Jin, X., Sun, D., Gao, H., *et al.* 2018. Five novel col7a1 gene mutations in three chinese patients with recessive dystrophic epidermolysis bullosa. *Ann Clin Lab Sci,* 48**;** 100-105.

Yasukawa, K., Sawamura, D., Goto, M., Nakamura, H., Jung, S. Y., Kim, S. C. dan Shimizu, H. 2006. Epidermolysis bullosa simplex in japanese and korean patients: Genetic studies in 19 cases. *Br J Dermatol,* 155**;** 313-317.

Ye, S., Yan, F., Wang, H., Mo, X., Liu, J., Zhang, Y., Li, H., *et al.* 2021. Diversity analysis of gut microbiota between healthy controls and those with atopic dermatitis in a chinese population. *J Dermatol,* 48**;** 158-167.

Yenamandra, V. K., Bhari, N., Ray, S. B., Sreenivas, V., Dinda, A. K., Scaria, V., Sharma, V. K., *et al.* 2017a. Diagnosis of inherited epidermolysis bullosa in resource-limited settings: Immunohistochemistry revisited. *Dermatology,* 233**;** 326-332.

Yenamandra, V. K., Moss, C., Sreenivas, V., Khan, M., Sivasubbu, S., Sharma, V. K. dan Sethuraman, G. 2017b. Development of a clinical diagnostic matrix for characterizing inherited epidermolysis bullosa. *Br J Dermatol,* 176**;** 1624-1632.

Yenamandra, V. K., Shamsudheen, K. V., Madhumita, R. C., Rijith, J., Ankit, V., Scaria, V., Sridhar, S., *et al.* 2017c. Autosomal recessive epidermolysis bullosa simplex: Report of three cases from india. *Clin Exp Dermatol,* 42**;** 800-803.

Yenamandra, V. K., Vellarikkal, S. K., Kumar, M., Chowdhury, M. R., Jayarajan, R., Verma, A., Scaria, V., *et al.* 2017d. Application of whole exome sequencing in elucidating the phenotype and genotype spectrum of junctional epidermolysis bullosa: A preliminary experience of a tertiary care centre in india. *J Dermatol Sci,* 86**;** 30-36.

UNIVERSITAS
GADJAH MADA
Variasi klinis, mutasi genetik, dan mikrobiom saluran cerna pada epidermolisis bulosa
SUCI WIDHIATI, Prof. Dr. Hardyanto Soebono, dr., SpKK(K)
Universitas Gadjah Mada, 2022 | Diunduh dari http://etd.repository.ugm.ac.id/

Zeeuwen, P. L., Kleerebezem, M., Timmerman, H. M. dan Schalkwijk, J. 2013. Microbiome and skin diseases. *Curr Opin Allergy Clin Immunol*, 13**;** 514-520.