

## Marco JANNER, MD

### 1. ORIGINAL ARTICLES

#### 2023

Christoph Saner, Alistair M. Senior, Hanyue Zhang, Aino-Maija Eloranta, Costan G. Magnussen, Matthew A. Sabin, Markus Juonala, Marco Janner, David P. Burgner, Ursula Schwab, Eero A. Haapala, Berit L. Heitmann, Stephen J. Simpson, David Raubenheimer and Timo A. Lakka. *Evidence for protein leverage in a general population sample of children and adolescents*. European Journal of Clinical Nutrition 2023; <https://doi.org/10.1038/s41430-023-01276-w>

#### 2022

Janner M, Saner C. *Impact of Type 1 Diabetes Mellitus on Bone Health in Children*. Horm Res Paediatr, 2022, doi: 10.1159/000521627.

#### 2021

Koch G, Steffens B, Leroux S, Gotta V, Schropp J, Gächter P, Bachmann F, Weizel T, Janner M, L'Allemand D, Konrad D, Szinnai G, Pfister M. *Modeling of levothyroxine in newborns and infants with congenital hypothyroidism: challenges and opportunities of a rare disease multi-center study*. J Pharmocokinet Pharmacodyn, 2021 Oct,48(5):711-723. doi: 10.1007/s10928-021-09765-w.

Saner C, Laitinen TT, Nuotio J, Arnup SJ, Harcourt BE, Bekkering S, McCallum Z, Kao KT, Janner M, Magnussen CG, Sabin MA, Juonala M, Burgner DP. *Modest decrease in severity of obesity in adolescence associates with low arterial stiffness*. Atherosclerosis 2021. DOI: 10.1016/j.atherosclerosis.2021.09.013

#### 2020

Marco Janner, Grit Sommer, Michael Groessl, Christa E. Flück. *Premature adrenarche in girls is characterized by enhanced 17,20-lyase and 17 $\beta$ -hydroxysteroid dehydrogenase activities*. Journal of Clinical Endocrinology and Metabolism 2020: <https://doi.org/10.1210/clinem/dgaa598>.

#### 2019

Shahida Moosa, Guilherme L. Yamamoto, Lutz Garbes, Katharina Keupp, Ana Belezza-Meireles, Carolina Araujo Moreno, Eugenia Ribeiro Valadares, Sergio B. de Sousa, Sofia Maia, Jorge Saraiva, Rachel S. Honjo, Chong Ae Kim, Hamilton Cabral de Menezes, Ekkehart Lausch, Pablo Villavicencio Lorini, Arsonval Lamounier, Jr., Tulio Canella Bezerra Carniero, Cecilia Giunta, Marianne Rohrbach, Marco Janner, Oliver Semler, Filippo Beleggia, Yun Li, Gökhan Yigit, Nadine Reintjes, Janine Altmüller, Peter Nürnberg, Denise P. Cavalcanti, Bernhard Zabel, Matthew L. Warman, Debora R. Bertola, Bernd Wollnik, Christian Netzer. *Autosomal-Recessive Mutations in MESD Cause Osteogenesis Imperfecta*. American J Hum Genet 2019:1-8. doi:10.1016/j.ajhg.2019.08.008.

M. Santi, M. Janner, G.D. Simonetti, S.A.G. Lava. *Prescription of vitamin D among Swiss pediatrician*. European journal of Pediatrics 2019;178(7):1119-23.

## 2015-2018

N. Marti, J. Malikova, J.A. Galván, M. Aebischer, M. Janner, Z. Sumnik, B. Obermannnova, G. Escher, A. Perren, C.E. Flück. *Androgen production in pediatric adrenocortical tumors may occur via both the classic and/or the alternative backdoor pathway*. [Mol Cell Endocrinol](#). 2017 May 10. pii: S0303-7207(17)30272-1. doi: 10.1016/j.mce.2017.05.014.

C. Saner, G.D. Giacometti, E. Wühl, P.E. Mullis, M Janner. *Circadian and ultradian cardiovascular rhythmicity in obese children*. Eur J Pediatr, doi:10.1007/s00431-016-2736-4.

M. C. Miletta, A. Eblé, M. Janner, S. Parween, A. V. Pandey, C. E. Flück, and P. E. Mullis. *IGHD II: a Novel GH-1 Gene Mutation (GH-L76P) Severely Affects GH-Folding, Stability and Secretion*. J Clin Endocrinol Metabol 2015, doi: 10.1210/jc.2015-3265 (early release).

L. Mansour-Hendili, Blanchard A, Le Pottier N, Roncelin I, Lourdel S, Treard C, González W, Vergara-Jaque A, Morin G, Colin E, Holder-Espinasse M, Bacchetta J, Baudouin V, Benoit S, Bérard E, Bourdat-Michel G, Bouchireb K, Burtey S, Cailliez M, Cardon G, Cartery C, Champion G, Chauveau D, Cochat P, Dahan K, De la Faille R, Debray FG, Dehoux L, Deschenes G, Desport E, Devuyt O, Dieguez S, Emma F, Fischbach M, Fouque D, Fourcade J, François H, Gilbert-Dussardier B, Hannedouche T, Houillier P, Izzedine H, Janner M, Karras A, Knebelmann B, Lavocat MP, Lemoine S, Leroy V, Loirat C, Macher MA, Martin-Coignard D, Morin D, Niaudet P, Nivet H, Nobili F, Novo R, Faivre L, Rigotherier C, Roussey-Kesler G, Salomon R, Schleich A, Sellier-Leclerc AL, Souлами K, Tiple A, Ulinski T, Vanhille P, Van Regemorter N, Jeunemaître X, Vargas-Poussou R.

*Mutation Update of the CLCN5 Gene Responsible for Dent Disease 1*. Hum Mut. 2015; 36(8):743-52.

C. Saner, G.D. Simonetti, E. Wühl, P.E. Mullis, M. Janner. *Ambulatory arterial stiffness index is increased in obese children*. Atherosclerosis (2015); 238:185-189.

M.-A. Burckhardt, Obmann V., Wolf R., Janner M., Flück C.E., Mullis P. *Ovarian and uterine development and hormonal feedback mechanism in a 46 XX patient with CYP19A1 deficiency under low dose estrogen replacement*. Gynecological Endocrinology 2015;31(5):394-54.

## 2009-2014

E. Del Pozo, M. Janner, A.R. Mackenzie et al. *Radiological, hormonal and biological correlates of skeletal growth in the female rat from birth to senescence*. Growth Horm IGF 1 Res. 2014; 24(2-3):83-8.

D. Dasgupta, M.J. Wee, M. Reyes, Y. Li, P.J. Simm, A. Sharma, K.-P. Schlingmann, M. Janner et al. *Mutations in SLC34A3/NPT2c Are Associated with Kidney Stones and Nephrocalcinosis*. J Am Soc Nephrol 2014; 25(10):2366-75.

A. Bieri, Oser-Meier M., Janner M., Cripe-Mamie C., Pipczynski-Suter K., Mullis P.E., Flück C.E. *Children's and adolescent's self - assessment of metabolic control versus professional judgment: a cross-sectional retrospective and prospective cohort study*. Int J Ped Endocr 2013(1): 21; doi: 10.1186/1687-9856-2013-21

N. Camats, Pandey AV, Fernández-Cancio M, Andaluz P, Janner M, Torán N, Moreno

F, Bereket A, Akcay T, García-García E, Muñoz MT, Gracia R, Nistal M, Castaño L, Mullis PE, Carrascosa A, Audí L, Flück CE. *Ten Novel Mutations in the NR5A1 Gene Cause Disordered Sex Development in 46,XY and Ovarian Insufficiency in 46,XX Individuals*. J Clin Endocrinol Metab (2012); 97(7): E1294-306.

M. Janner, Flück C.E., Mullis P.E.: *Impact of Estrogen Replacement throughout Childhood on Growth, Pituitary-Gonadal Axis and Bone in a 46,XX Patient with CYP19A1 deficiency*. Horm Res Paediatr 2012 (doi: 10.1159/000341585).

M.Janner, P. Ballinari, P.E. Mullis and C.E. Flück. *High prevalence of vitamin D3 deficiency in children and adolescents with type 1 diabetes*. Swiss Med Weekly (2010) 140:E1.

E. Del Pozo, Zapf J, Mackenzie AR, Janner M, Perrelet R, Lippuner K, Mullis P. *Experimental arthritis: Effect on growth parameters and total skeletal calcium*. Growth Horm IGF Res. 2009 (19):442-6.

## 2000-2008

B.Räz\*, M. Janner\*, V. Petkovic, D. Lochmatter, A. Eblée, M.T. Dattani, P.C. Hindmarsh, C.E. Flück and P.E. Mullis. *Growth response and final height in patients with severe GH deficiency: Does the d3- or full-length GH receptor isoform really matter?* J Clin Endocrinol Metab. 2008 Mar;93(3):974-80.

M. Janner, A.V. Pandey, P.E. Mullis and C. E. Flueck. *Clinical and biochemical description of a novel CYP21A2 gene mutation 962\_963insA using a new 3D model for the P450c21 protein*. Eur J Endocrinol (2006); 155:143-151.

M. Janner, P.E. Mullis. *Osteopenie und pathologische Frakturen bei einem Jugendlichen mit Laktoseintoleranz und hohem Oxalatkonsum*. Monatsschrift Kinderheilkunde (2005);153:360-3.

## 1991-1999

M. Janner, S.E. Knill, P. Diem, K.A. Zuppinger, P.E. Mullis. *Persistent microalbuminuria in adolescents with type I diabetes mellitus is associated to early rather than late puberty. Results of a prospective longitudinal study*. Eur J Ped (1994); 153:403-408.

M. Janner, R. C. Mühlbauer and H. Fleisch. *Sodium EDTA enhances intestinal absorption of two bisphosphonates*. Calcif Tissue Int (1991); 49:280-283.

R.C. Mühlbauer, F. Bauss, R. Schenk, M. Janner, E. Bosies, K. Strein and H. Fleisch. *BM 21.0955, a potent new bisphosphonate to inhibit bone resorption*. J Bone Min Res (1991); 6:1003-1011.

## 2. REVIEW ARTICLES

Janner M, Saner C. *The Impact of Type 1 Diabetes Mellitus on Bone Health in Children*. Horm Res Paediatr, 2021 Dec 22. DOI: 10.1159/000521627. Online ahead of print.

L. Bonafé, C. Giunta, C. Hasler, M. Janner, M. Kränzlin, B. Link, C. Meier, L.E. Ramseier, M. Rohrbach, S. Unger. *Osteogenesis imperfecta: Klinik, Diagnose und Management vom Kindes- bis ins Erwachsenenalter*. Swiss Med Wkly (2013); 46:925-931.

D. L'Allemand, T. Neuhaus, M. Janner, C. Brägger, J. Laimbacher. *Empfehlungen des Bundesamtes für Gesundheit zur Vitamin-D-Versorgung in der Schweiz – was bedeuten sie für den Pädiater?* Paediatrica (2012); 23(4):22-24.

B. Molinari-Büchi, J. Barth, M. Janner, P. Frey. *Surcharge pondérale et obésité chez l'enfant: les acquis et les nouvelles tendances.* Rev Méd Suisse (2010);249:1022-5.

P.E. Mullis, M. Janner. *Das Wachstum, ein zentraler Prozess, Teil 1.* Schweiz. Med. Forum (2009);33:560-565.

P.E. Mullis, M. Janner. *Das Wachstum, ein zentraler Prozess, Teil 2.* Schweiz. Med. Forum (2009);34:587-592.

M. Janner, P.E. Mullis. *Störungen der Kalziumhomöostase im Kindesalter – wann muss man daran denken?* Ther Umschau (2007); 64(5):271-275.

M. Janner, P.E. Mullis, C.E. Flück. *Das metabolische Syndrom: eine neue Kinderkrankheit?* Praxis (2006);95:493-500.

M. Janner. *Die XXL Generation. Eine neue Herausforderung im 21. Jahrhundert.* Paediatrica (2005); 16(4):40-43.

### 3. BOOK CHAPTERS

*Atlas der Entwicklungsdiagnostik.* Hsg Thomas Baumann. 2. Auflage 2007, Georg Thieme Verlag, Stuttgart: 71-92.

*Atlas der Entwicklungsdiagnostik.* Hsg Thomas Baumann. 3. Auflage 2013, Georg Thieme Verlag, Stuttgart: 288-317, 321-329.