

## Gemeinschaftspraxis für Humangenetik



Dr. med. Andrea Bier  
Priv.-Doz. Dr. med. Stefan Krüger, MHBA  
Dr. med. Silke Reif

Gutenbergstraße 5 · 01307 Dresden  
Tel. 0351 / 44 66 34 0  
Fax 0351 / 44 66 34 15  
praxis@medizinische-genetik-dresden.de  
www.medizinische-genetik-dresden.de

**Fachärzte für Humangenetik**

### Publikationen, umgekehrt chronologisch, Stand 31.01.2024

(kumulativ **405,018** Impact-Punkte aus **98** bewerteten Publikationen)

212. Mutational spectrum of suspected Charcot-Marie-Tooth disease in 399 patients. **Klaschka V, Großmann M, Bier A, Reif S, Timmer M, Jänecke C, Tchertov J, Kuchler B, Plaschke J, Krüger S.** Abstractband 34. gfh-Jahrestagung 2023:254.
211. Shifting the border of pathogenicity with a DEL9 mutation in the CEL gene. **Grossmann M, Wilk J, Bier A, Reif S, Timmer M, Jänecke C, Klaschka V, Kuchler B, Kobelt A, Krüger S, Plaschke J.** European Journal of Human Genetics (2023) 31:435.
210. A rare case of 3MC syndrome with thoracic aortic dissection. **Grossmann M, Krüger S, Bier A, Reif S, Jänecke C, Klaschka V, Plaschke J, Weiss N, Abicht A, Timmer M.** Abstractband 32. gfh-Jahrestagung 2023:125.
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207. Axenfeld-Rieger Anomaly and Neuropsychiatric Problems-More than Meets the Eye. Safari A, Ziegler A, Merkschlager A, **Krüger S**, Kölker S, Hoffmann GF, Syrbe S. *Neuropediatrics*. 2020 Jun;51(3):192-197. [Impact Factor 1,530]
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203. Painful Charcot-Marie-Tooth neuropathy type 2E/1F due to a novel NEFL mutation. Doppler K, Kunstmann E, **Krüger S**, Sommer C. *Muscle Nerve*. 2017 May;55(5):752-755. [Impact Factor 2,713]
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