

## 2025

### **Accurate Diagnosis of Congenital Adrenal Hyperplasia due to CYP21A2 Variants Requires Promoter Analysis.**

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### **Aktualisierung des Modells pathogener CNBP Repeatexpansionen ursächlich für die Myotone Dystrophie Typ 2.**

Erdmann H, Wendlandt M, Rost S, Lucas MC, Sauer M, Golibruch M, Cordts I, Kleefeld F, Timmer M, Bier A, Wunderlich G, Becker K, Kleinle S, Wenninger S, Walter M, Neuhann T, Holinski-Feder E, Schoser B, Abicht A.

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<https://pubmed.ncbi.nlm.nih.gov/36693418/>

### **Parallel in-depth analysis of repeat expansions in ataxia patients by long-read sequencing.**

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### **Closing the Gap - Detection of 5q-Spinal Muscular Atrophy by Short-Read Next-Generation Sequencing and Unexpected Results in a Diagnostic Patient Cohort.**

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