

Summary Advances in ADH1 Diagnosis and Treatment



Emphasize the need for early, accurate diagnosis: genetic testing

Commercial Testing	Focus	Clinical Relevance
<u>PreventionGenetics</u>	HypoPara testing	Free CASR-related testing via Calcilytix; 26 genes included, expands access to diagnosis.
<u>Ambry Genetics</u>	Exome & disease-specific panels	Broad test menu; useful for endocrine and metabolic disorders.
<u>Invitae Panel</u>	Hypoparathyroidism gene panel	Includes CASR, GCM2, and related genes; supports differential diagnosis.
<u>Sequencing.com</u>	Genome data tools & apps	Helps interpret raw DNA data (e.g., 23andMe) for health insights; HIPAA-compliant.