

Disease

Aromatic amino acid decarboxylase deficiency^{1, 2, 3}

Gene

DDC

Inheritance	AR
Current Treatments	Pyridoxine/pyridoxal phosphate, folinic acid, dopamine agonists, MAO B inhibitors. Gene therapy PTC-AADC
Evidence for Treatment	guideline ⁴
Non-Molecular Testing Availability	yes
Non-Molecular Test	urine organic acid analysis, plasma AADC activity ⁵
Disease Frequency (per 100,000)	0.83 ⁶
Genes Included in Disease Frequency	DDC
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Aromatic+Amino+Acid+Decarboxylase+Deficiency&term=&cntry

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/31975548>
- ² <https://pubmed.ncbi.nlm.nih.gov/37803205/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/34253733/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/28100251/>
- ⁵ <https://www.sciencedirect.com/science/article/abs/pii/S1096719220301517>
- ⁶ <https://pubmed.ncbi.nlm.nih.gov/30952622/>