Rx-Genes
Treatment Guidance at the Point of Care

Disease

Tuberous sclerosis 1[1, 2]

Gene

TSC1

Inheritance	AD
Current Treatments	Vigabatrin for seizures, mTOR inhibitor for rhabdomyoma, Epidiolex (cannabidiol) for seizures
Evidence for Treatment	guideline[3, 4]
Non-Molecular Testing Availability	no
ClinGen	https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC026
Disease Frequency (per 100,000)	10.205[5]
Genes Included in Disease Frequency	TSC1 and TSC2
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Tuberous+Sclerosis&term=&cntry=&state=&city=&dist=

References

1 https://www.ncbi.nlm.nih.gov/books/NBK1220/

2 https://www.ncbi.nlm.nih.gov/pubmed/29759095

3 https://pubmed.ncbi.nlm.nih.gov/31039793/

4 https://pubmed.ncbi.nlm.nih.gov/30247655/

5 https://www.ncbi.nlm.nih.gov/pubmed/30016967