



# General Genetics CPC

## Evidence Statement

The Clinical Prioritisation Criteria (CPC) were created using a comprehensive consultation, review, and feedback process. Clinical Pathway Group (CPG) members employed a standardised yet adaptable methodology to complete the CPC's development. This included the selection of clinical leads through self-nomination or nomination by peers.

The CPG comprised of representatives from various healthcare fields, including clinicians, general practitioners, allied health professionals, and nurses. The CPC development was informed by a variety of sources, such as current clinical practice, research, existing guidelines, as well as relevant SA Health policies.

CPC development was informed by:

- existing local, national and international guidelines and/or criteria (refer to references below)
- evidence based practice recommendations
- SA Health policy guidelines
- national definitions for elective surgery urgency categories (AIHW)

Where possible, the CPC represents current clinical consensus about criteria for accessing specialist care and treatment to public specialist outpatient services in South Australia. This CPC will be reviewed and updated every 2 years, to ensure they are reflective of contemporary practices.

### Reference List

1. Australian Genetic Heart Disease Registry 2012, *Familial Dilated Cardiomyopathy (FDC or DCM)*, updated December 12 2012, viewed February 3 2023, <[http://www.heartregistry.org.au/wp-content/uploads/2012/12/AGHDR\\_DCMInfoSheet\\_2012.pdf](http://www.heartregistry.org.au/wp-content/uploads/2012/12/AGHDR_DCMInfoSheet_2012.pdf)>.
2. Australian Genetic Heart Disease Registry 2012, *Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)*, updated December 6 2012, viewed February 3 2023, <<http://www.heartregistry.org.au/patients-families/genetic-heart-diseases/arrhythmogenic-right-ventricular-cardiomyopathy/>>.
3. Australian Genetic Heart Disease Registry 2012, *Long QT Syndrome (LQTS)*, updated December 10 2012, viewed February 3 2023, <[http://www.heartregistry.org.au/wp-content/uploads/2012/12/AGHDR\\_LQTSInfoSheet\\_2012.pdf](http://www.heartregistry.org.au/wp-content/uploads/2012/12/AGHDR_LQTSInfoSheet_2012.pdf)>.
4. Australian Genetic Heart Disease Registry 2012, *Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)*, updated December 12 2012, viewed February 3 2023, <[http://www.heartregistry.org.au/wp-content/uploads/2012/12/AGHDR\\_CPVTInfoSheet\\_2012.pdf](http://www.heartregistry.org.au/wp-content/uploads/2012/12/AGHDR_CPVTInfoSheet_2012.pdf)>.
5. Australian Genetic Heart Disease Registry 2012, *Left Ventricular Noncompaction (LVNC)*, updated December 12 2012, viewed February 3 2023, <<http://www.heartregistry.org.au/patients-families/genetic-heart-diseases/left-ventricular-noncompaction-lvnc/>>.
6. Australian Genetic Heart Disease Registry 2014, *Brugada Syndrome*, updated September 3 2014, viewed February 3 2023, <<http://www.heartregistry.org.au/patients-families/genetic-heart-diseases/brugada-syndrome/>>.

# General Genetics CPC

## Evidence Statement



7. Australian Genetic Heart Disease Registry 2016, *Hypertrophic Cardiomyopathy (HCM)*, updated January 17 2016, viewed February 2 2023, <<http://www.heartregistry.org.au/patients-families/genetic-heart-diseases/hypertrophic-cardiomyopathy/>>.
8. Brett, T, Radford, J, Heal, C, Hespe, C, Garton-Smith, J, Kirke, A, . . . Watts, G 2021, 'Implications of new clinical practice guidance on familial hypercholesterolaemia for Australian general practitioners', *Australian Journal for General Practitioners*, vol. 50, 08/23, pp. 616-621.
9. Cardiac Inherited Diseases Group 2023, *Cardiac Inherited Diseases*, viewed February 3 2023, <<https://www.cidg.org.nz/patients-families/cardiac-inherited-diseases/>>.
10. Cardiac Inherited Diseases Group 2023, *Health Professionals*, viewed February 3 2023, <<https://www.cidg.org.nz/health-professionals/>>.
11. Cardiac Inherited Diseases Group 2023, *Patients & Families - Cardiac Inherited Diseases*, viewed 13 February 2023, <<https://www.cidg.org.nz/patients-families/cardiac-inherited-diseases/>>.
12. Cardiac Society of Australia and New Zealand 2023, *Position Statements and Practice Guidelines - Cardiovascular Genetics*, viewed February 3 2023, <<https://www.csanz.edu.au/for-professionals/position-statements-and-practice-guidelines/#cardiovascular-genetics>>.
13. Central Adelaide Local Health Network 2022, *CALHN Adult Genetics Unit - Landing page*, SA Health, <<https://www.sahealth.sa.gov.au/wps/wcm/connect/public+content/sa+health+internet/services/hospitals/outpatient+services/outpatient+clinics/central+adelaide+lhnspecialist+and+outpatient+clinics/adult+genetics+unit+outpatient+service+in+calhn>>.
14. Central Adelaide Local Health Network 2023, *Endocrine and metabolic outpatient service in CALHN*, viewed 20 March 2023, <<https://www.sahealth.sa.gov.au/wps/wcm/connect/public+content/sa+health+internet/services/hospitals/outpatient+services/outpatient+clinics/central+adelaide+lhnspecialist+and+outpatient+clinics/endocrine+and+metabolic+outpatient+service+in+calhn>>.
15. Ehlers-Danlos Support UK 2017, *The Ehlers-Danlos syndromes (EDS) GP Toolkit*, viewed February 13 2023, <<https://gptoolkit.ehlers-danlos.org/>>.
16. EndoGen 2023, *EndoGen Online Resources*, viewed 13 February 2023, <<https://www.endogenaustralia.com/>>.
17. Health Centre for Genetics Education 2022, *MTHFR Gene Testing*, NSW Government, viewed 8 December 2022, <[https://www.genetics.edu.au/PDF/MTHFR\\_gene\\_testing\\_fact\\_sheet-CGE.pdf](https://www.genetics.edu.au/PDF/MTHFR_gene_testing_fact_sheet-CGE.pdf)>.
18. Health Centre for Genetics Education 2022, *Fact Sheet quicklinks*, NSW Government, viewed 8 December 2022, <<https://www.genetics.edu.au/SitePages/A-Z-fact-sheets.aspx>>.
19. Health Direct 2020, *Guide to Genetic Disorders*, viewed January 5 2023, <<https://www.healthdirect.gov.au/guide-to-genetic-disorders>>.

# General Genetics CPC

## Evidence Statement



20. Hickey, SE, Curry, CJ & Toriello, HV 2013, 'American College of Medical Genetics and Genomics
21. Practice Guideline: lack of evidence for MTHFR polymorphism testing', *Genet Med*, vol. 15, no. 2, Feb, pp. 153-156.
22. Human Genetics Society of Australasia 2023, *HGSA Consumer Links*, viewed January 5 2023, <<https://www.hgsa.org.au/Web/Web/Consumer-resources/Consumer-resources.aspx?hkey=e6e6f1f5-8f34-4fbb-b6e6-a829d799e932>>.
23. Human Genetics Society of Australasia 2023, *HGSA Provider Resources*, viewed January 5 2023, <<https://www.hgsa.org.au/Web/Web/HP-Resources/HP-Resources.aspx?hkey=7a0e5949-07cb-4d51-beb6-3b4925f64d5c>>.
24. Huntington's Disease News 2023, *Huntington's Disease News*, viewed February 13 2023, <<https://huntingtonsdiseasenews.com/>>.
25. Huntington's Disease Society of America 2023, *The Huntington's Disease Coalition for Patient Engagement (HD-COPE)*, viewed February 13 2023, <<https://hdsa.org/hd-research/hd-cope/>>.
26. Huntington's Disease Youth Organization 2023, *Huntington's Disease Youth Organization - HDYO*, viewed 20 March 2023, <<https://en.hdyo.org/>>.
27. Huntington's South Australia & Northern Territory Inc 2018, *Huntington's South Australia & Northern Territory Inc*, viewed 20 March 2023, <<https://huntingtonssant.org.au/>>.
28. Kidney Health Australia 2020, *Kidney Health Australia*, viewed 20 March 2023, <<https://kidney.org.au/>>.
29. MedlinePlus 2023, *Genetics*, viewed 20 March 2023, <<https://medlineplus.gov/genetics/>>.
30. Metro North Health Genomic Institute 2022, *The Genetic Kidney Disease Decision Aids*, Queensland Government, viewed 20 March 2023, <<https://metronorth.health.qld.gov.au/genomic-institute/clinical-care/gkdda>>.
31. National Health Service 2021, *Huntington's Disease*, updated March 29, 2021, viewed February 13 2023, <<https://www.nhs.uk/conditions/huntingtons-disease/>>.
32. National Health Service Bristol 2022, *Alpha-1 antitrypsin fact sheet for Primary Care*, 3 edn, updated November 15 2022, viewed February 3 2023, <[https://www.nbt.nhs.uk/sites/default/files/document/Alpha-1%20antitrypsin%20fact%20sheet%20for%20primary%20care\\_0.pdf](https://www.nbt.nhs.uk/sites/default/files/document/Alpha-1%20antitrypsin%20fact%20sheet%20for%20primary%20care_0.pdf)>.
33. Northern Sydney Local Health District 2023, *Clinical Genetics Clinician Resources*, New South Wales Government viewed January 5 2023, <<https://www.nslhd.health.nsw.gov.au/genetics/Pages/Clinician-Resources.aspx#referral>>.
34. NSW Centre for Genetics Education 2021, *Chromosome microarray (CMA) testing in children and adults*, viewed 20 March 2023, <<https://www.genetics.edu.au/SitePages/Chromosome-microarray-fact-sheet.aspx>>.
35. NSW Centre for Genetics Education 2015, *Hereditary Haemochromatosis - Fact Sheet*, updated December 11 2015, viewed February 3 2023, <<https://www.genetics.edu.au/SitePages/Hereditary-haemochromatosis.aspx>>.

# General Genetics CPC

## Evidence Statement



36. NSW Health Centre for Genetics Education 2018, *Huntington disease*, viewed 20 March 2023, <<https://www.genetics.edu.au/SitePages/Huntington-disease.aspx>>.
37. Ommen, SR, Mital, S, Burke, MA, Day, SM, Deswal, A, Elliott, P, . . . Sorajja, P 2020, '2020 AHA/ACC Guideline for the Diagnosis and Treatment of Patients With Hypertrophic Cardiomyopathy: A Report of the American College of Cardiology/American Heart Association Joint Committee on Clinical Practice Guidelines', *Circulation*, vol. 142, no. 25, Dec 22, pp. e558-e631.
38. Pathology Tests Explained 2022, *Alpha-1-antitrypsin*, updated 22 May 2022, Australasian Association for Clinical Biochemistry and Laboratory Medicine, viewed February 2 2023, <<https://pathologytestsexplained.org.au/learning/test-index/alpha1-antitrypsin>>.
39. QIMR Berghofer Medical Research Institute 2020, *Your Blood, Your Story*, viewed 20 March 2023, <<https://www.youtube.com/watch?v=Z3mQCcb6oas>>.
40. Queensland Health 2022, *Clinical Prioritisation Criteria - Genetics - Cardiac genetics*, Queensland Health, <<https://cpc.health.qld.gov.au/Condition/300/cardiac-genetics-general-genetics>>.
41. Queensland Health 2022, *Clinical Prioritisation Criteria - Genetics - Endocrine genetics*, Queensland Health, <<https://cpc.health.qld.gov.au/Condition/302/endocrine-genetics-general-genetics>>.
42. Queensland Health 2022, *Clinical Prioritisation Criteria - Genetics - Gastroenterology/Hepatology genetics*, Queensland Health, <<https://cpc.health.qld.gov.au/Condition/304/gastroenterology-hepatology-genetics-general>>.
43. Queensland Health 2022, *Clinical Prioritisation Criteria - Genetics - Metabolic genetics*, Queensland Health, <<https://cpc.health.qld.gov.au/Condition/306/metabolic-genetics-general-genetics>>.
44. Queensland Health 2022, *Clinical Prioritisation Criteria - Genetics - Neurology genetics*, Queensland Health, <<https://cpc.health.qld.gov.au/Condition/307/neurology-genetics-general-genetics>>.
45. Queensland Health 2022, *Clinical Prioritisation Criteria - Genetics - Ophthalmic genetics*, Queensland Health, <<https://cpc.health.qld.gov.au/Condition/309/ophthalmic-genetics-general-genetics>>.
46. Queensland Health 2022, *Clinical Prioritisation Criteria - Genetics - Other adult genetic conditions*, Queensland Health, <<https://cpc.health.qld.gov.au/Condition/327/other-adult-genetic-conditions>>.
47. Queensland Health 2022, *Clinical Prioritisation Criteria - Genetics - Paediatric genetics*, Queensland Health, <<https://cpc.health.qld.gov.au/Condition/321/paediatric-genetics>>.
48. Queensland Health 2022, *Clinical Prioritisation Criteria - Genetics - Renal Genetics*, Queensland Health, <<https://cpc.health.qld.gov.au/Condition/322/renal-genetics-adultpaediatric>>.



# General Genetics CPC

## Evidence Statement



49. Queensland Health 2022, *Clinical Prioritisation Criteria - Genetics - Reproductive genetics*, Queensland Health, <<https://cpc.health.qld.gov.au/Condition/325/reproductive-genetics>>.
50. Queensland Health 2022, *Clinical Prioritisation Criteria - Genetics - Respiratory Genetics*, Queensland Health, <<https://cpc.health.qld.gov.au/Condition/326/respiratory-genetics-adultspaediatric>>.
51. Queensland Health 2022, *Clinical Prioritisation Criteria - Genetics - Untested and AFFECTED blood relative of a person with an identified gene mutation/chromosomal anomaly (confirmatory testing)*, Queensland Health, <<https://cpc.health.qld.gov.au/Condition/323/untested-and-affected-blood-relative-of-a-per>>.
52. Queensland Health 2022, *Clinical Prioritisation Criteria - Genetics - Untested and UNAFFECTED blood relative of a person with an identified gene mutation/chromosomal anomaly (predictive/presymptomatic testing)*, Queensland Health, <<https://cpc.health.qld.gov.au/Condition/324/untested-and-unaaffected-blood-relative-of-a-p>>.
53. Retina Australia 2023, *Inherited Retinal Disease*, viewed 20 March 2023, <<https://retinaaustralia.com.au/>>.
54. SA Pathology 2023, *Maternal and Genetic Carrier Screening*, viewed January 5 2023, <<https://www.sapathology.sa.gov.au/wps/wcm/connect/SA+Pathology+Internet+Content+New/Content/Clinicians/Maternal+and+Genetic+Carrier+Screening/>>.
55. Southern Adelaide Local Health Network 2023, *SA Huntington's Disease Service - Landing page Flinders Medical Centre*, viewed February 13 2023, <<https://www.sahealth.sa.gov.au/wps/wcm/connect/public+content/sa+health+internet/services/hospitals/flinders+medical+centre/services+and+clinics+at+flinders+medical+centre/services+at+flinders+medical+centre/huntingtons+disease+service+sa+at+flinders+medical+centre>>.
56. The National Health Service 2018, *National genomic test directory*, updated 31 October 2022, viewed 8 December 2022, <<https://www.england.nhs.uk/publication/national-genomic-test-directories/>>.
57. The Royal Australian College of General Practitioners 2018, *Genomics in General Practice*, updated 1 December 2022, Melbourne, Vic, viewed 8 December 2022, <<https://www.racgp.org.au/clinical-resources/clinical-guidelines/key-racgp-guidelines/view-all-racgp-guidelines/genomics-in-general-practice/disease-specific-topics/mthfr-gene-testing>>.
58. The Royal Australian College of General Practitioners 2018, *Disease specific topics - Haemoglobinopathies*, 2nd edn, viewed 13 February 2023, <<https://www.racgp.org.au/clinical-resources/clinical-guidelines/key-racgp-guidelines/view-all-racgp-guidelines/genomics-in-general-practice/disease-specific-topics/haemoglobinopathies>>.
59. The Royal Australian College of General Practitioners 2021, *Genetic counselling and testing*, updated December 12 2021, viewed January 5 2023, <<https://www.racgp.org.au/clinical-resources/clinical-guidelines/key-racgp-guidelines/view->>



# General Genetics CPC

## Evidence Statement

[all-racgp-guidelines/guidelines-for-preventive-activities-in-general-pr/genetic-counselling-and-testing](#)>.

60. Unique - Understanding Rare Chromosome and Gene Disorders 2020, *Rare Chromosome & Gene Disorder Guides*, updated October 2, 2020, viewed January 5 2023, <<https://rarechromo.org/disorder-guides/>>.
61. University of Exeter Medical School & Royal Devon University Healthcare NHS Foundation Trust 2023, *Diabetes Genes*, DiabetesGenes, viewed February 3 2023, <<https://www.diabetesgenes.org/>>.
62. University of Exeter Medical School & Royal Devon University Healthcare NHS Foundation Trust 2023, *MODY Probability Calculator*, Exeter Diabetes, viewed 13 February 2023, <<https://www.diabetesgenes.org/exeter-diabetes-app/ModyCalculator>>.
63. Vears, DF, Ayres, S, Boyle, J, Mansour, J & Newson, AJ 2020, 'Human Genetics Society of Australasia Position Statement: Predictive and Presymptomatic Genetic Testing in Adults and Children', *Twin Research and Human Genetics*, vol. 23, no. 3, pp. 184-189.
64. Women's and Children's Health Network 2022, *SA Clinical Genetics Service - Landing page WCHN*, SA Health, <<https://www.wch.sa.gov.au/patients-visitors/screening-testing-prevention/sa-clinical-genetics-service>>.