SDH-related familial paraganglioma-phaeochromocytoma – risk management

ID: 3558 v.3

Related pages:

- Informing family members about hereditary cancer
- Cancer predisposition genes: population carrier frequency
- Paraganglioma-phaeochromocytoma – panel testing
- Facts for people and families with a faulty SDHA gene
- Facts for people and families with a faulty SDHB gene
- Facts for people and families with a faulty SDHC gene
- Facts for people and families with a faulty SDHD gene
- Facts for people and families with a faulty SDHAF2 gene

History

Version 3

Date	Summary of changes
18/12/2019	"Mutation" changed to "pathogenic variant" throughout document for consistency among eviQ cancer genetics protocols per agreement among the cancer genetics reference committees' chairs. Definition of "pathogenic variant" added as a pop-up. Protocol version number increased to V.3.
18/12/2020	The following sections of the protocol were updated to align with the revised eviQ cancer genetics risk management template: • Lifetime risk of cancer: risk for general population updated to include AIHW 2016 data • Cancer risk management guidelines: ◦ Sentence added above table "The choice of risk management strategy should take into account current age, other health issues and residual cancer risk." ◦ Other table formatting changes • Support and information: template wording updated.

Version 2

Date	Summary of changes
15/04/2019	New protocol presented at Nov 2018 reference committee meeting. Discussion continued over email and approved for publication. • Protocol replaces protocols ID 752 and ID 1278 which have now been archived. • Protocol approved as V.1. To be reviewed in 2 years.
28/08/2019	Protocol title changed from 'Risk management for SDH-related paraganglioma-phaeochromocytoma predisposition syndromes' to 'SDH-related familial paraganglioma-phaeochromocytoma – risk management' in accordance with Cancer Genetics Reference Committees' consensus. Version number increased to V.2.

As ID 3558 replaces two existing approved protocols, their individual history sections are included below for consistency in documentation.

ID 750 Risk management for paraganglioma-phaeochromocytoma predisposition syndromes (SDHA, SDHB and SDHC gene mutations)

Date	Summary of changes
20/12/2010	Approved.

31/05/2012	Discussed at national reference committee meeting April 2012 and decision made for this protocol to cover the following gene mutations SDHA, SDHB and SDHC only. SDHD and SDHAF2 to be covered by a separate protocol. The following changes have been made under: • **Title** of protocol changed to reflect gene mutations covered SDHA, SDHB and SDHC. • **Target group -** SDHA gene mutation added. • **Exculsion criteria -** SDHAF2 mutation and other paraganglioma/phaeochromocytoma predisposition genes added RET and TMEM127 added. • **Lifetime risk of cancer table -** incidence of tumour development for SDHB mutation updated and SDHD mutation removed. • **Cancer risk management guidelines** updated to include specific recommendations for phaeochormocytoma and paraganglioma surveillance and prevention for SDHB, SDHA and SDHC. • **Evidence for risk management guidelines** - paraganglioma/phaeochromocytoma Surveillance - some of the information is based on expert opinion. • Evidence for gastric GIST added. • **Reference List** - updated and additional references moved to History tab. For access to previous version please contact us if required.
10/06/2014	Discussed at October 30, 2013 reference committee meeting, discussion continued via email and the following changes made: • **Exclusion criteria** - updated. • **Lifetime risk of cancer** - table updated. • **Cancer risk management guidelines** - table - surveillance for SDHB and for all SDHA and SDHC updated. Prevention for SDHA, SDHB and SDHC - updated. • **Evidence for risk management guidelines** - paraganglioma/phaeochromocytoma - surveillance updated. renal - updated. • **Support and information**- updated. • **Reference list -** references reviewed and updated, further references added to history. • For review second yearly.
31/07/2015	Link to AGSA changed to Genetic Alliance Australia.
07/01/2016	Sentence added to Risk Management template: "The impact of lifestyle on cancer risk should be discussed".
14/04/2016	Sentence added to Risk Management template: "This risk management guideline has been developed for individuals who have NOT been diagnosed with a relevant cancer/tumour. The care of affected individuals should be individualised based on their clinical situation, and the monitoring they need as part of their treatment and post-treatment follow up".
31/05/2017	Transferred to new eviQ website. Version number changed to V.4.
15/04/2019	Protocol archived, as reviewed prior to Nov 2018 reference committee meeting and replaced by protocol ID 3558 - Risk management for SDH-related paraganglioma-phaeochromocytoma predisposition syndromes.
	Further references used to develop this protocol: 1. Hensen, E. F. and J. P. Bayley. 2011. "Recent advances in the genetics of SDH-related paraganglioma and pheochromocytoma." Fam Cancer 10(2):355-363. 2. Schiavi, F., R. L. Milne, E. Anda, et al. 2010. "Are we overestimating the penetrance of mutations in SDHB?" Hum Mutat 31(6):761-762 3. Neumann, H. P., C. Pawlu, M. Peczkowska, et al. 2004. "Distinct clinical features of paraganglioma syndromes associated with SDHB and SDHD gene mutations." JAMA 292(8):943-951. 4. Benn, D. E., A. P. Gimenez-Roqueplo, J. R. Reilly, et al. 2006. "Clinical presentation and penetrance of pheochromocytoma/paraganglioma syndromes." J Clin Endocrinol Metab 91(3):827-836 5. Ricketts, C. J., J. R. Forman, E. Rattenberry, et al. 2010. "Tumor risks and genotype-phenotype-proteotype analysis in 358 patients with germline mutations in SDHB and SDHD." Hum Mutat 31(1):41-51 6. Lemaire, M., A. Persu, P. Hainaut, et al. 1999. "Hereditary paraganglioma." J Intern Med 246(1):113-116 7. Pasini, B., S. R. McWhinney, T. Bei, et al. 2008. "Clinical and molecular genetics of patients with the Carney-Stratakis syndrome and germline mutations of the genes coding for the succinate dehydrogenase subunits

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15. Dundee, P., B. Clancy, S. Wagstaff, et al. 2005. "Paraganglioma: the role of genetic counselling and radiological screening." J Clin Neurosci 12(4):464-466

16. Havekes, B., K. King, E. W. Lai, J. A. Romijn, E. P. Corssmit and K. Pacak. 2010. "New imaging approaches to phaeochromocytomas and paragangliomas." Clin Endocrinol (Oxf) 72(2):137-145

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ID 1278 Risk management for paraganglioma-phaeochromocytoma predisposition syndromes (SDHD and SDHAF2 gene mutations)

Date	Summary of changes
29/05/2012	Discussed at national reference committee meeting April 2012 and decision made to develop this protocol for

	SDHD and SDHAF2 gene mutations. **Evidence for risk management guidelines** - Some of the information is based on expert opinion. Approved.
05/12/2013	**Cancer risk management guidelines - phaeochromocytoma and paraganglioma - Surveillance -** typographical error noted between these dates 29 May to 5 December 2012, screening for MRI shown as 2 yearly but should be 3 yearly. Therefore corrected as below: • **from age 10:** recommend 3 yearly MRI of base of skull to coccyx include kidneys and adrenals
10/06/2014	Discussed at October 30 2013 reference committee meeting, discussion continued via email and the following changes made: • **Exclusion criteria** - updated. • **Lifetime risk of cancer** - paternally inherited mutations and table updated. • **cancer risk management guidelines** - table - surveillance and prevention updated for phaeochromocytoma & paraganglioma • **Evidence for risk management guidelines** - paraganglioma/phaeochromocytoma - surveillance updated renal - updated parent of origin effect updated. • **Support and information** - updated. • **Reference list** - references reviewed and updated, further references added to history. • For review second yearly.
31/07/2015	Link to AGSA changed to Genetic Alliance Australia.
07/01/2016	Sentence added to Risk Management template: the impact of lifestyle on cancer risk should be discussed.
14/04/2016	Sentence added to Risk Management template: "This risk management guideline has been developed for individuals who have NOT been diagnosed with a relevant cancer/tumour. The care of affected individuals should be individualised based on their clinical situation, and the monitoring they need as part of their treatment and post-treatment follow up".
31/05/2017	Transferred to new eviQ website. Version number changed to V.3.
15/04/2019	Protocol archived ,as reviewed prior to Nov 2018 reference committee meeting and replaced by protocol ID 3558 - Risk management for SDH-related paraganglioma-phaeochromocytoma predisposition syndromes.
	Further references used to develop this protocol: 1. Hensen, E. F. and J. P. Bayley. 2011. "Recent advances in the genetics of SDH-related paraganglioma and pheochromocytoma." Fam Cancer 10(2):355-363 2. Neumann, H. P., C. Pawlu, M. Peczkowska, et al. 2004. "Distinct clinical features of paraganglioma syndromes associated with SDHB and SDHD gene mutations." JAMA 292(8):943-951. 3. Benn, D. E., A. P. Gimenez-Roqueplo, J. R. Reilly, et al. 2006. "Clinical presentation and penetrance of pheochromocytoma/paraganglioma syndromes." J Clin Endocrinol Metab 91(3):827-836 4. Ricketts, C. J., J. R. Forman, E. Rattenberry, et al. 2010. "Tumor risks and genotype-phenotype-proteotype analysis in 358 patients with germline mutations in SDHB and SDHD." Hum Mutat 31(1):41-51 5. Hensen, E. F., J. C. Jansen, M. D. Siemers, et al. 2010. "The Dutch founder mutation SDHD.D92Y shows a reduced penetrance for the development of paragangliomas in a large multigenerational family." Eur J Hum Genet 18(1):62-66 6. Lemaire, M., A. Persu, P. Hainaut, et al. 1999. "Hereditary paraganglioma." J Intern Med 246(1):113-116 7. Pasini, B., S. R. McWhinney, T. Bei, et al. 2008. "Clinical and molecular genetics of patients with the Carney-Stratakis syndrome and germline mutations of the genes coding for the succinate dehydrogenase subunits SDHB, SDHC, and SDHD." Eur J Hum Genet 16(1):79-88 8. Gill, A. J., A. Chou, R. E. Vilain, et al. 2011. ""Pediatric-type" gastrointestinal stromal tumors are SDHB negative ("type 2") GISTs." Am J Surg Pathol 35(8):1245-1247; author reply 1247-1248 9. Havekes, B., A. A. van der Klaauw, M. M. Weiss, et al. 2009. "Pheochromocytomas and extra-adrenal paragangliomas detected by screening in patients with SDHD-associated head-and-neck paragangliomas."

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