

SDHD-related head and neck paragangliomas & their natural course Heesterman, B.L.

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ABBREVIATIONS

α-**KG** *α*-ketoglutarate

A Adenine
Arg Arginine
Asp Aspartic acid

AUC Area under the curve

bp Base pairC Cytosine

c. Coding DNA reference sequence

CA California

CBT Carotid body tumor

CDKN1C Cyclin-dependent kinase inhibitor 1c

CI Confidence interval

cm Centimeter

CNS Central nervous system
CSF Cerebrospinal fluid
CT Computed tomography
DCIS Ductal carcinoma in situ

del Deletion

df Degrees of freedom **DNA** Deoxyribonucleic acid

DSA Digital substraction angiography3D TOF MRA 3D Time of Flight MR Angriography

ECA External carotid artery
EGLN Elegans homolog
ENT Ear nose and throat

EPAS₁ Endothelial PAS domain protein ₁

EPO Erytropoetin

ERK Extracellular signal-regulated kinases

FAD Flavin adenine dinucleotide

¹⁸**FDOPA** ¹⁸F-fluordopa

Fe Iron

FH Fumurate hydratase

G Guanine ⁶⁸**Ga** ⁶⁸Gallium

GIST Gastrointestinal stromal tumor

GN Ganglioneuroma

GPR91 G-protein-coupled receptor 91

HIF Hypoxia-inducible factor

HNPGL Head and neck paraganglioma

HR Hazard ratio

HRE HIF-responsive elements

IBM International Business Machines

ICA Internal carotid artery

Ile Isoleucine

¹²³I-MIBG ¹²³I-metaiodobenzylguanidine

IQR Interquartile range

KIF1B β Kinesin family member 1B β

LC Lung carcinoma

LDGA Laboratory for Diagnostic Genome Analysis

Leu Leucine

LOVD Leiden Open (source) Variation Database

LUMC Leiden University Medical Center

MAX Myc associated factor X

5mC 5-methylcytosine

MDH2 Malate dehydrogenase 2

MERTK C-MER proto-oncogene tyrosine kinase

Met Methionine

MIBG Metaiodobenzylguanidine

MLPA Multiplex ligation-dependent probe amplification

mm Millimeter

MRA Magnetic resonance angiography

MRI Magnetic resonance imaging

ms Millisecond

3MT 3-methoxytyramine

MTC Medullary thyroid carcinoma
mTOR Mammalian target of rapamycin

n Number

NVII Facial nerve

NIX Glossopharyngeal nerve

NX Vagus nerve

NXII Hypoglossal nerve
NB Neuroblastoma

NF1 Neurofibromatosis type 1
NGF Nerve growth factor

NGS Next generation sequencing

NG_ Genomic sequence

NM_ mRNA reference sequence
NT_ DNA reference sequence

NY New York
OR Odds ratio

p. Protein sequencePA Pituitary adenomaPCC Pheochromocytoma

PET Positron emission tomography

PGL Paraganglioma

PHDs Prolyl hydroxylase domain proteins

PNMT Phenylethanolamine N-methyltransferase

PP Predicted probability

Pro Proline

PTC Papillary thyroid carcinoma

Q Quartile

r Growth rate

R² Coefficient of determination

RCC Renal cell carcinoma

RET Rearranged during transfection proto-oncogene

RMSE Root mean squared error

ROC Receiver operating characteristic

ROS Reactive oxygen species

S Sulfur

SD Standard deviation

SDD Smallest detectable difference
SDH Succinate dehydrogenase

SDHA Succinate dehydrogenase subunit-A (flavoprotein-subunit)

SDHAF1 Succinate dehydrogenase, assembly factor 1
SDHAF2 Succinate dehydrogenase, assembly factor 2

SDHB Succinate dehydrogenase subunit-B (iron-sulfur subunit)
SDHC Succinate dehydrogenase subunit-C (anchoring subunit)
SDHD Succinate dehydrogenase subunit-D (anchoring subunit)

SLC22A18 poly-specific organic cation transporter

SMR Standardized mortality ratio

sPGLExtra-adrenal sympathetic paragangliomaSPSSStatistical Package for the Social Sciences

T Thymine
T Tesla
t Time

TCATricarboxylic acid T_d Tumor doubling time

Thr Threonine

TMEM127 Transmembrane protein 127

TX Texas
Tyr Tyrosine

USA United States of America

VEGF Vascular endothelial growth factor

VHL Von Hippel-Lindau

V Volume

WHO World health organization

LIST OF CONTRIBUTING AUTHORS

J.P. Bayley, PhD

Department of Human Genetics, Leiden University Medical Center

Prof. P.P.G. van Benthem, MD, PhD

Department of Otorhinolaryngology, Leiden University Medical Center

B.T.J. van Brussel

Department of Clinical Genetics, Leiden University Medical Center

J.M. Bokhorst, BSc

Eindhoven University of Technology

E.P.M. Corssmit, MD, PhD

Department of Endocrinology, Leiden University Medical Center

Prof. O.M. Dekkers, MD, PhD

Department of Endocrinology & Department of Epidemiology, Leiden University Medical Center

Prof. J.F. Hamming, MD, PhD

Department of Surgery, Leiden University Medical Center

L.T. van Hulsteijn, MD, PhD

Department of Endocrinology, Leiden University Medical Center

F.J. Hes, MD, PhD

Department of Clinical Genetics, Leiden University Medical Center

J.C. Jansen, MD, PhD

Department of Otorhinolaryngology, Leiden University Medical Center

A.G.L. van der Mey, MD, PhD

Department of Otorhinolaryngology, Leiden University Medical Center

L.H.M. de Pont, BSc

Department of Otorhinolaryngology, Leiden University Medical Center

C.M.J. Tops, PhD

Department of Clinical Genetics, Leiden University Medical Center

B.M. Verbist, MD, PhD

Department of Radiology, Leiden University Medical Center

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- **B. L. Heesterman**, J. P. Bayley, C. M. Tops, et al. "High prevalence of occult paragangliomas in asymptomatic carriers of SDHD and SDHB gene mutations." In: Eur. J. Hum. Genet. 21.4 (Apr. 2013), pp. 469–70.
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- **B. L. Heesterman**, L.M.H. de Pont, B.M. Verbist, et al. "Age and tumor volume predict growth of carotid and vagal body paragangliomas." In: J Neurol Surg B Skull Base. 78.6 (Dec. 2017), pp. 497–505.
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- **B. L. Heesterman**, J. Bokhorst, L.M.H. de Pont, et al. "Mathematical models for tumor growth and the reduction of overtreatment." In: J Neurol Surg B Skull Base. (in press).

ABOUT THE AUTHOR

Berdine Louise Heesterman was born on March 3, 1990 in Baarn, the Netherlands. She completed secondary school at "Het Baarnsch Lyceum" in 2008, after which she started studying medicine at the University of Leiden. During her studies she began her research at the department of Otorhinolaryngology with which she continued after obtaining her medical degree in 2015. Currently she is working as a consultant at IG&H, primarily within the healthcare sector.