

SUPPLEMENTARY TABLE S1

A case report of syndromic multinodular goitre in adolescence:

Exploring the phenotypic overlap between Cowden and DICER1 syndromes

Dorothee Bouron-Dal Soglio, MD, PhD¹, **Leanne de Kock**, B-Tech^{2,3}, **Richard Gauci**, MBBS, FRACP⁷, **Nelly Sabbaghian**, MSc², **Elizabeth Thomas**, MBBS (Hons), FRACP^{7,8}, **Helen C. Atkinson**, BSc(Hons) PhD⁹, **Nicholas Pachter**, MBChB, FRACP^{9,10}, **Simon Ryan**, MBBS, FRACS¹³, **John P. Walsh**, MBBS, FRACP, PhD^{5,9}, **M. Priyanthi Kumarasinghe**, MBBS, MD, FRCPA, DipCytopathol(RCPA)⁴, **Karen Carpenter**, BSc, PhD, FHGSA¹⁰, **Ayça Aydoğan**⁶, **Colin J.R. Stewart**, MBChB, FRCPath, FRCPA⁹, **William D. Foulkes**, MBBS, PhD^{2,3,12}, **Catherine S. Choong**, MBBS, MD, FRACP^{8,9}

Supplementary Table S1: Cowden syndrome diagnostic criteria

Major Criteria
Adult-onset Lhermitte-Duclos disease
Mucocutaneous lesion (e.g. facial trichilemmomas, acral keratoses, papillomatous lesions)
Breast carcinoma
Thyroid carcinoma (papillary or follicular)
Macrocephaly > 97 th percentile
Endometrial carcinoma
Minor Criteria
Other thyroid lesions (multinodular goitre, adenoma)
Mental retardation
Gastrointestinal hamartoma
Fibrocystic lesions of the breast
Lipomas
Fibromas
Genitourinary tumours or malformations

Reference:

Hobert JA, Eng C 2009 PTEN hamartoma tumor syndrome: an overview. Genetics in medicine 11:687-694.