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CASE REPORT

Cowden syndrome and reconstructive breast surgery: Case reports and review of the literature

E. Ali ^a, P.G. Athanasopoulos ^b, P. Forouhi ^a, C.M. Malata ^{a,b,*}

^a Cambridge Breast Unit, Addenbrooke's University Hospital, Cambridge, UK

^b Department of Plastic & Reconstructive Surgery, Addenbrooke's University Hospital, Cambridge, UK

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Expandable implants

Summary Cowden syndrome (CS) is a rare, autosomal dominant inherited disorder associated with multiple benign and malignant neoplasms, including breast cancer. Prophylactic resection of susceptible organs remains controversial.

We briefly describe the syndrome and review management of the risk of CS-associated malignancies. Three cases of bilateral risk-reducing mastectomy and immediate breast reconstruction are described.

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The phosphatase and tensin (*PTEN*) homologue on chromosome ten is a tumour suppressor gene involved in the PI3K/Akt/mTOR pathway which regulates several critical cellular functions including cell proliferation, apoptosis and migration.¹ A germline mutation in the *PTEN* gene results in Cowden syndrome (CS) (Table 1a). CS has a Caucasian and female predominance² and a prevalence of 1/200,000

population.³ Many cases are considered unrecognised due to the variable phenotype of the disease⁴ so this figure is likely to be an underestimate.

CS is associated with an increased risk of developing neoplasia of tissues derived from all three embryonic germ cell layers, especially of the breast, thyroid and endometrium.² Management options range from observation and screening, to surgery for diagnosed cancer. The lifetime risk associated with the development of thyroid cancer, mostly follicular, is up to 10% in both genders compared to <1% in the general population. Endometrial cancer, mainly adenocarcinoma, has an estimated lifetime risk of up to 10% compared with <2.5% in the general population. The lifetime risk of developing breast cancer for a woman

* Corresponding author at: Department of Plastic and Reconstructive Surgery, Addenbrooke's Hospital, Cambridge University Hospitals NHS Foundation Trust, Cambridge, CB2 2QQ, United Kingdom. Tel.: +44 1223 586672; fax: +44 1223 257177.

E-mail address: cmalata@hotmail.com (C.M. Malata).

Table 1a The pathognomonic, major and minor features of Cowden Syndrome according to US National Comprehensive Cancer Network (NCCN) Genetics/High Risk Panel operational diagnostic criteria for 2008.

Pathognomonic features	Major features	Minor features
<ul style="list-style-type: none"> • Lhermitte-Duclos disease • Mucocutaneous neoplasms (trichilemmomas, acral keratoses, verucoid or papillomatous papules) 	<ul style="list-style-type: none"> • Breast cancer • Follicular or papillary thyroid cancer • Macrocephaly • Endometrial cancer 	<ul style="list-style-type: none"> • Structural thyroid lesions e.g. adenomas or multinodular goitres • Mental retardation with IQ \leq 75 • Gastro-intestinal tract hamartomas • Fibrocystic breast disease • Lipomas • Fibromas • Genitourinary tumours or structural malformations • Uterine fibroids

Table 1b Summary of the underlying breast disease and the breast reconstruction performed in each of the three cases.

Case number	Patient age, years	Site of lesion and result(s) of core biopsy	Breast cup size	Breast surgery performed	Breast reconstruction surgery performed
1	43	No breast lesions were identified	38C	Bilateral skin sparing mastectomy only removing the nipple	Immediate expander reconstruction: Natrelle (formerly McGhan), Style 150 short height expandable implants, Inamed Aesthetic Ltd., Co. Wicklow, Ireland
2	25	Right sided phyllodes tumour with associated intra duct lobular neoplasia Left sided atypical hyperplasia (Non-malignant lesions associated with an increased risk of developing breast cancer)	32B	Bilateral skin sparing mastectomy only removing the nipple	Immediate expander reconstruction: Expander as above
3	47	No breast lesions were identified	36DD	Bilateral skin sparing mastectomy only removing the nipple	DIEP and SIEA flaps: Deep inferior epigastric perforator Superficial inferior epigastric artery

Table 1c Summary of the characteristic features of Cowden Syndrome demonstrated in each of the three cases.

Case	Macrocephaly	GIT disease	Thyroid disease	Genitourinary disease	Other lesions
1	Present	Gastric, duodenal and colonic polyps	Previous thyroidectomy for benign nodular goitre	Previous risk-reducing hysterectomy	- Hamartomatous tongue nodules - Previous malignant melanoma
2	Present	Not present	Previous thyroidectomy for a papillary tumour of unknown malignant potential	Normal vaginal US/S and hysteroscopy	- Hamartomatous tongue nodules
3	Present	Not present	Previous thyroidectomy for follicular carcinoma	Previous hysterectomy for uterine fibroids	Not present

US/S, ultra sound scan.

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