Cate’s Story:
Hereditary Diffuse Gastric Cancer

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Cate is a 25-year-old woman who has a germline mutation in *cadherin 1* (*CDH1*), coupled with a significant family history of diffuse gastric cancer. Cate’s journey started with her seeing her mother and sister die at a young age from this disease. Rather than being daunted by this adversity, she showed great resilience, coping with not only her grief but also simultaneously dealing with the impact of finding that she was a carrier of the same genetic disorder.

Gastric Cancer

Gastric cancer is a major cause of cancer-related mortality worldwide (Black, Kaneshiro, Lai, & Shimizu, 2014) and is thought to be responsible for about 10% of cancer-related deaths across the globe (Worster et al., 2014). Although it is a significant problem in the Western world, it is far more common in Asia (Macdonald, 2011), where it is a significant public health burden (Tan & Wong, 2012).

A small proportion of all gastric cancers (1%–3%) arise because of a known hereditary syndrome, the most common of which is hereditary diffuse gastric cancer (HDGC) (Pattison & Boussioutas, 2015). This is an autosomal dominant genetic disease characterized by an increased risk of developing diffuse gastric cancer at a young age. The gene responsible for HDGC is *CDH1*, also known as *E-cadherin*, a germline mutation conferring an 80% risk of developing gastric cancer during the lifetime of the carrier (Black et al., 2014, Pattison & Boussioutas, 2015). Overt gastric cancer frequently appears before 40 years of age but also has been reported in teenage *CDH1* carriers (Framp, 2010). Females with germline *CDH1* mutations face an additional risk of developing lobular breast cancer, with a reported cumulative risk of 60% by the age of 80 years (Pattison & Boussioutas, 2015).

**CDH1 Mutation Carriers**

HDGC is characterized by high disease penetrance, and the management of identified carriers is complicated by the lack of a reliable surveillance method for effective early detection and diagnosis of the multiple signet ring cell carcinomas that are seen with HDGC. Routine endoscopy for known *CDH1* mutation carriers has proven to be ineffective in the setting of a cancer that is typically subtle, infiltrating beneath normal mucosa and leaving the stomach to appear normal (Pattison & Boussioutas, 2015).

Genetic Testing

Cate’s mother and sister died of gastric cancer at ages 30 and 22 years, respectively. Before she died, Cate’s sister had genetic testing and was identified as a carrier for the *CDH1* gene mutation. Cate’s sister is the index patient in this family because the identification of her genetic mutation raised the question of genetic testing for other relatives, including Cate, who was her only sibling (Fitzgerald & Caldas, 2006). Cate’s mother came from a large family, and this diagnosis and its implications naturally generated much anxiety. The emotional impact of the diagnosis of a genetic disorder such as this cannot be underestimated and can have a huge impact on family members, forcing them to make difficult decisions about their future (Lynch et al., 2008; Mahon 2014). Given the clinical and psychological implications of testing positive as a carrier for a genetic disorder, at-risk individuals must receive counselling with a genetics service before any testing takes place (Fitzgerald & Caldas, 2006).

Management

*CDH1* mutation carriers, such as Cate, should be managed by a team of healthcare professionals (Pattison & Boussioutas, 2015; van der Post et al., 2015). Prior to coming to the multidisciplinary clinic at the Peter MacCallum Cancer Centre in Melbourne, Australia, Cate attended a familial cancer center for genetic counseling and subsequent mutation testing. She demonstrated good insight regarding the implications of having a *CDH1* mutation. She was made aware of the statistics around her lifetime risk of developing gastric

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adenocarcinoma and that endoscopic surveillance is not sensitive enough to detect early malignant tumor growth. Having seen her mother and sister die at a young age, she was keen to pursue risk-reduction surgery. At the cancer center, she met a specialist upper gastrointestinal surgeon, dietitian, and nurse experienced in this area. She had extensive preoperative discussions regarding the principles of laparoscopic prophylactic gastrectomy, including potential perioperative complications, as well as potential functional sequelae postoperatively. On reflection, Cate said she felt well informed and prepared for surgery but felt she did not completely comprehend some potential symptoms, particularly dumping syndrome, until she actually experienced them.

Psychosocial Assessment

Cate was seen some months before her planned surgery because young CDH1 mutation carriers with no evidence of malignancy have the luxury of time to prepare themselves. Psychosocial assessment revealed that she was seeing a psychologist for preexisting bereavement issues, which she found helpful, but she was interested in referral to a social worker for additional counseling and advice around sickness benefits because she did not have any paid sick leave entitlement. She described having supportive friends but also feeling disconnected at times, believing they could not understand what she was about to go through.

Feelings of isolation led Cate to start writing a blog to share her experiences and help others in her position; she also hoped that it may help raise awareness about this rare type of cancer. The blog provided a forum for her “bucket list.” The list was started after encouragement from her friends after the initial diagnosis when she was feeling very lost. The overseas trip she started planning before her surgery motivated her to keep moving forward, particularly when times were challenging in the immediate postoperative period and in the first few weeks after discharge.

Cate described writing her blog as a coping strategy because she did not know anyone else who had this genetic mutation or who had undertaken this particular type of surgery. In the time since she began writing about her experience, carriers of CDH1 mutations have connected with her via the Internet. Her story can be found on the No Stomach for Cancer website (http://beginningnottheend.blogspot.com.au).

Cate also credits her writing for helping her move on from initial thoughts of anger and self-pity. Now, she said she feels positive because she has realized that she has been given a chance at a long life that was not afforded to her mother or sister.

Prophylactic Surgery

Cate had an uneventful laparoscopic total gastrectomy with Roux-en-Y jejunal reconstruction and was discharged from the hospital five days later. Histo logic examination of the surgical specimen revealed two tiny foci of signet ring adenocarcinoma situated close to the posterior body of the stomach. A focus of in situ signet ring cell adenocarcinoma was also seen in the posterior fundus, but 16 regional nodes showed no evidence of involvement by carcinoma. Finding sites of cancer is not an uncommon scenario in the pathology specimens of CDH1 carriers, like Cate, who proceed to prophylactic gastrectomy having previously had negative gastroscopy results (Lynch et al., 2008; Pattison & Bousioutas, 2015). This may act as affirmation that patients at high risk have made the right decision to have prophylactic surgery.

Postoperative Period

Although Cate’s postoperative period was relatively uneventful, she recalled struggling with some symptoms after surgery. Initially, she was unable to drink adequate amounts of fluid. She also experienced problems with constipation and associated bloating. Cate had discomfort for the first 10 days after discharge and some occasional symptoms of dumping syndrome. Such issues are to be expected postgastroctomy, and, for Cate, these symptoms were short-lived. She was back working full-time three months after surgery. At five months postsurgery, she had gained 7 kg, resumed all normal activities, and was about to leave on her long-awaited trip overseas. A study by van der Post et al. (2015) showed that quality-of-life indicators return to preoperative levels about 12 months after surgery, despite lingering symptoms for some patients.

In the time since Cate’s surgery, many of her relatives have undergone genetic counselling and testing. A number have been found to be carriers of the CDH1 gene mutation and have gone on to have the same risk-reduction surgery at the cancer center. Previously published family case histories have reported that not all individuals who are confirmed as CDH1 mutation carriers will proceed to have risk-reduction surgery (Lynch et al., 2008). Some are concerned about implications for their work, and others report the idea of the surgery as being too extreme. Within many families, some do not even proceed to counselling and testing because they do not want to know.

Conclusion

CDH1 mutation carriers represent a unique group of previsors, or survivors who have a genetic predisposition for developing cancer (Mahon, 2014). These patients are considerably younger than the average person who has a gastrectomy for sporadic gastric cancer, and the long-term sequelae could be different (Pattison & Bousioutas, 2015). This remains an area in need of additional studies to improve patient management. Understanding is also lacking of the considerable long-term psychological challenges for these patients, their families, and their children (Mahon, 2014; Wilcox, Perpich, Noffsinger, Posner, & Cooper, 2011).

Cate’s story demonstrates how an individual who is otherwise healthy chose a life-changing strategy to reduce the risk of developing cancer and adapted well physically and emotionally. She has been an avid contributor to web-based information and a support blog and allowed others to follow her personal story. She also has undertaken a television interview. Her personal resilience and capacity to adapt, together with her willingness to share her story, have enabled her to raise awareness of this rare type of cancer and also support and inspire others, in the hope.
that they do not feel so alone in their journey.

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References


Do You Have an Interesting Topic to Share?

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