Paving the Way: A Grounded Theory of Discovery and Decision-Making for Persons Diagnosed With the CDH1 Marker

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Abstract

Purpose: To understand the process of discovery and decision-making for adults with the CDH1 marker for hereditary diffuse gastric cancer (HDGC) and inherited breast cancer.

Participants and Setting: Purposeful sampling included 20 participants; 17 adults (11 women and 6 men, ages 23–77) recruited through the No Stomach for Cancer organization; six participants were interviewed two times;with three healthcare providers also interviewed. Nineteen interviews were by telephone; one was in person.

Methodological Approach: Grounded theory with constant comparison.

Findings: The person diagnosed with the genetic marker CDH1 undergoes the decision-making process of *Paving the way* as they address this healthcare challenge. *Paving the way* explains the entry points for learning the risk, discerning testing for confirmation, choosing iterative individual cycles of surveillance, surgery, and ongoing adjustments postoperatively while normalizing to live longer.

Implications for Nursing: Understanding the process of *Paving the way* explains and describes the nine key factors for decision-making and predicts the timing for nursing interventions for both post-genetic testing and pre- and postoperative assessment and planning.

Knowledge Translation: Advocacy for the self and family is key to *Paving the Way*. Nursing has an opportunity to develop and expand the roles for navigator and counselor in the area of genetic testing. Patients undergoing PTG have chronic healthcare needs. Family implications for genetic testing require assessment beyond the individual.

Keywords: CDH1, grounded theory, genetics, gastric cancer, breast cancer.

Stomach cancer is the fifth leading cause of cancer worldwide with 1,033,701 estimated new cases and 782,685 deaths reported in 2018 (Bray, et al., 2018). The survival rate is particularly poor (4%) for those diagnosed with advanced disease (No Stomach for Cancer [NSFC], 2019; Pernot et al., 2015). Inherited forms of gastrointestinal cancer (GC) occur in approximately 1–3% of adults. Hereditary diffuse gastric cancer (HDGC) is a rare form of GC that is difficult to detect and has a very poor prognosis (van der Post et al., 2015). HDGC is a germline (inheritable) cell-to-cell adhesion protein e-cadherin gene—known as CDH1—first discovered by a team of researchers within a native Maori family in New Zealand (Guilford et al., 1999) and since identified in Europe, Canada, and the United.States (U.S.). (Corso, Marrelli, Pascale, Vindigni, & Roviello, 2012). The risk for CDH1-positive persons of developing HDGC is 70% (95% CI, 59–80) for men by age 80 and 56% (95% CI, 44–69) for women and, additionally, for women a 42% (95% CI, 23–68) risk for lobular breast cancer (BC; Kaurah & Huntsman, 2018). Statistical data for CDH1-related male BC remains undetermined. The average age of HDGC at diagnosis is 38; however, it can occur in adolescents and adults at ages 14–69 (Hansford et al., 2015; van der Post et al., 2015). Detection of HDGC is difficult due to the insidious growth of the tumor, which begins underneath the lining of the stomach in poorly differentiated signet ring cell cancer (Onitilo, Aryal, & Engel, 2013; Pernot et al., 2015) and the lack of observable presenting symptoms in these patients (Hebbard et al., 2009; Mastoraki et al., 2011; van der Post et al., 2015).

Once a positive blood test for the CDH1 marker is determined, prophylactic total gastrectomy (PTG) is the recommended treatment to prevent aggressive adenocarcinoma (van der Post et al., 2015). CDH1 testing is also recommended for family members (van der Post et al.,

2015). This recommendation results in additional decision-making issues related to family disclosure, genetic testing, and further screening.

The CDH1-positive person, whether male or female, faces difficult decisions: (a) electing surveillance with endoscopy, (b) removing the entire stomach to avoid gastric cancer, (c) screening for BC, and/or (d) choosing a prophylactic double mastectomy to avoid BC (Hallowell, Badger, et al., 2016; Hallowell, Lawton, et al., 2016; Hallowell et al., 2017).

Information is limited for how CDH1-positive persons embark on the decision-making process. Explicating this process could inform interventions for (a) clinical practice, (b) creating decision-making aids, and (c) providing needed patient and family support (Schreiber & Stern, 2001). Therefore, the purpose of this study was to understand the process of discovery and decision-making for CDHI-positive adults regarding HDGC and inherited BC.

Methods

Participants and Setting

The grounded theory (GT) study was conducted over one year after obtaining IRB approval at the study site. Purposive sampling was used to recruit participants (N = 20). Inclusion criteria were as follows: English-speaking CDHI-positive adults, family members at risk for the marker (tested or not tested), and healthcare providers (HCPs) caring for CDH1-positive persons. Participants were recruited from the No Stomach for Cancer (NSFC, 2019) nonprofit organization, whose members total over 20,000, of whom a small percentage were positive for the CDH1 marker. Recruitment occurred via the NSFC website connected to the closed NSFC Facebook site by posting a study invitation, researcher contact information, and an IRB-approved fact sheet. Nineteen telephone and one in-person audio-recorded interviews were conducted with recorded verbal consent using a semiflexible interview guide and a demographic

form. Per GT theoretical sampling, six participants (33.3%) were interviewed a second time 4–6 months after the initial interview to further inform the substantive process.

Methodological Approach

The GT method by Glaser and Strauss (1967) and Glaser (2008) was used to develop the substantive theory. After consent, interviews lasted 30–90 minutes and were audio recorded. The semiflexible interview guide was modified and expanded in subsequent interviews as the data informing the substantive theory emerged.

Data management and analysis used classic GT constant comparison of the data by Glaser and Strauss and Glaser (1967, 2008. First-level, open, selective, and theoretical coding, with memo sorting were employed for the theoretical model, which included theoretical sampling to increase the depth and variation (Glaser, 2008; Schreiber & Stern, 2001). Theoretical saturation was reached when no new information emerged as categories and their properties developed. The theory was created by the principal investigator from the dense verbatim data and the variations that explained and encircled the social and behavioral descriptions provided by the participants. As the theory developed, peer debriefing, various drafts, and figures of the process were reviewed with the committee members.

Trustworthiness

Trustworthiness was maintained by prolonged engagement within the interviews, observing at the NSFC meetings, and by second interviews of 50% of the participants. Triangulation of the data sources was accomplished by the NSFC website, by later reviewing other oncology-related grounded theories, and by constant comparison of interviews of the HCPs, family members, and participant interviews. Data integrity was maintained with an audit trail,

field notes, and reflective writing (Glaser & Strauss, 2008; Lincoln & Guba, 1985). A member check was conducted with a 15-member review panel at the NSFC national meeting.

The final figure of the model (see Figure 1) was reviewed additionally with two HCPs for goodness of fit, workability, making sense, and understanding.

Findings

Paving the way describes a decision-making process that leads to living longer (see Figure 1). *Paving the way* is initiated to avoid cancer to live longer through a four-stage process beginning with *learning the risk of genetic susceptibility*, *discerning genetic susceptibility*, then iteratively *choosing interventions* to *normalizing* to live longer. Nine factors were identified during the study: Age, Advocacy, Cancer found, Emotions, Gastric consequences, HCPs, Life events, Resources, and Witnessing (see Figure 2). These factors influenced the variation in the behavior of the participants throughout the process.

Demographic and clinical characteristics are displayed in Table 1. The participants' mean age was 50 years (range 23–77), with a majority being female (65%), White (90%), and married (58.8%). Over forty percent (41.1%) had a first-generation positive CDH1 marker. Surveillance biopsies or surgical mapping biopsies were reported. Of the 14 participants with PTG, n=7 (50%) were biopsied while under routine surveillance at least once prior to surgery, and n=7 (50%) underwent only endoscopic mapping surveillance biopsy prior to surgery. After PTG surgery, 92.85% of the 14 PTG participants were found to have in situ (73.3%) or more advanced (13%) HDGC.

Stage 1

The first stage began with *learning of genetic susceptibility* for the CDH1 marker. Disclosure of risk for the marker was routed through three points of discovery: cancer found first

(lobular or HDGC) , family members, or referral to a genetic counselor with or without knowledge of the risk for the marker (see Table 2; Stage 1).

Cancer found first: The first point of entry occurred when *cancer is found.* A cancer diagnosis of the gastric tract or breast(s) was reported by four participants. As one stated, "I discovered that I had two lumps on one of my breasts… And a week later I had biopsy results come back that I had lobular BC." Another participant shared, "I was being followed for esophagitis, and I had a 1-year follow-up, and on the biopsies, there were a couple of cells that looked like signet ring cell adenocarcinoma." The cancer diagnosis along with the findings of lobular or signet ring cell cancer, a strong family history of cancer, or premenopausal BC prompted the HCP to order additional testing. The cancer found first entry point was rapid since participants were engaged in cancer treatment from the beginning.

Family member: The second entry point appeared when the genetic mutation risk was communicated to participants by *family members.* Nine participants (4 CDH1-positive persons and 5 family members) encountered this entry point. Mothers and fathers had conducted family meetings with their children, while others reached out to siblings, and still others to more distant relatives. Many family members shared their own testing results, informed participants of their risk, and encouraged acting on this new knowledge. However, not all family members passed along or shared news with others; e.g., an elderly man who was overwhelmed by another serious family illness disengaged from sharing this new information. Another participant reported a family member ignoring HCP instructions to share the news with family members. When a CDH1-positive person is under age 30, parents often lead the communication.

Genetic counselor: The third and last entry point of discovery occurred through the recommendation of an HCP or through self-referral to a genetic counselor. Participants in this

entry point were curious to know why their family history of cancer was so great. As one participant shared, “I have a strong family history of cancer. My mother was diagnosed with lobular BC and died before the age of 50.” Participants were referred based on a history of cancer through an HCP or by their own self-advocacy, insisting on a referral and testing related to a strong family history or a personal diagnosis of cancer. One participant’s rationale included “I just thought it was a good measure to take, just to make sure and find out where the lobular came from, no stone left unturned.”

Stage 2

Stage 2 began after learning the risk for the gene (see Table 2; Stage 2) in deciding to test or not for the gene.. Reactions to *learning of genetic susceptibility* were described as “unnerving,” “surprising,” and “unexpected.” For others, learning of the risk was consciously “denied” initially or persistently due to life events, like a serious family illness or an uneasiness about facing the risk for HDGC.

Participants at this stage sought resources themselves or learned from others through peer-to-peer resources. A frequent source of information was the Internet. One participant shared that after speaking to the genetic counselor, “I googled it all weekend!” Participants also used these resources: HCPs, genetic counselors, family members, especially parents, nonprofit organizations like NSFC, reading peer-reviewed journal articles, and tapping into blogs and instant messaging of others who were CDH1-positive. Factors that influenced the testing decision included cancer already present, the psychological impact of not knowing, fear of cancer, curiosity regarding a strong family or personal history of cancer, and family-planning concerns for the next generation.

Various reasons motivated participants to test. For some, by witnessing family members with cancer: “I saw what my cousin had gone through”—or self-symptoms: “I was having heartburn for about a year”—or concern regarding a lobular BC diagnosis: “I asked for genetic testing.” After a positive CDH1 marker test was confirmed, a myriad of new emotions arose, ranging from denial: “I didn’t believe it, I didn’t understand it,” or “It’s not part of me”—or a delayed response: “Later that day it actually hit me.” For some, the emotions ranged from “a huge relief” as it answered their question, to an extreme negative feeling: “I felt that I had been given a death sentence.” However, not everyone decided to test for the marker. A middle-aged family member who chose not to test explained it this way: “I block it out of my life.”

Once the decision was made, the speed for completing testing ranged from immediate to delayed. Self-advocacy was a key factor for testing referrals. Conditions for rapid testing included that cancer was already present, family-advocacy, and discerning reliable resources, such as the NSFC website and vetted social media sites. For some participants, the need to know the results of their personal risk for the gene was urgent due to concern for their children: “Well, I have two young boys, and I thought they have a 50/50 chance of getting it so I thought I’ve got to get it done.” For others the need for more rapid testing was due to the ending of insurance benefits or, as the endoscopic specialist shared, from a fear manifested in self-awareness of gastrointestinal symptoms. Alternatively, testing was delayed for others (see Table 3). Results of the testing were acknowledged immediately or questioned, the latter requiring a second confirmatory test as in the case of a participant who had not known of CDH1 in the family: “I paid for a test and had it, so I had it confirmed through a different testing company.” Two participants elected a second test for confirmation, while most did not. A genetic counselor shared the observation that the news of genetic confirmation for the CDH1 gene was an

extremely hard diagnosis to hear. Results were received by participants with various emotional responses from a delayed reaction to immediately laughing out loud or crying. Some reacted with a sense of panic, some with roiling anxiety, while others with quiet matter-of-fact acceptance. Emotional responses were varied: “I felt like I was hit with a Mack truck” or “I was doing fine until later that evening when it hit me”; when two siblings tested together: “Both my kids busted out laughing. They weren’t expecting both to be positive.” For those diagnosed with BC first, treatment for the cancer occurred with testing for all but one participant . After the testing results were known, the next step was *choosing interventions* to address the genetic risk (see Table 2; Stage 3).

Stage 3

Stage 3 began with *choosing interventions*, including surveillance, surgery, or treatment after PTG, and/or prophylactic mastectomy (see Table 4 and Figure 3). *Choosing interventions* was individually based on the factors underlying each person’s situation at the time of discovery. The point of discovery influenced the order of procedures chosen; e.g., when BC was diagnosed first in a participant, treatment continued and was not interrupted by a positive test for the gene. However, some surveillance for HDGC or lobular BC might have begun prior to surgery. As one participant said:

> I have been doing the mammogram, and I am getting an MRI. I'm looking to next year to have the bilateral mastectomy, and that is a depressing factor. I mean, it's like you lose part of what makes you feel a woman.

Emotions influenced engaging in next steps for *choosing interventions* were varied as family members were considered: “I had brought this into our family and now they would have

to deal with it for the rest of their lives"—and fears surfaced about the diagnosis: "It's so horrible and it's so evil and it's so frightening"—or the impact of witnessing others dying: "I have seen very young family members die from this and it was ugly and they were very, very sick"—to temporary denial: "The genetic counselor couldn't possibly be saying that I have to have my stomach removed." Two participants doubting their results retested and were informed of the same results with the second testing

Choosing interventions included, PTG, esophagogastroduoendoscopy (EGD), or breast surveillance, or chemotherapy. Treatments were influenced by age, self/family advocacy, cancer found, generational concerns, HCPs, psychological impact, resources for decision-making, and witnessing. Participants chose interventions sequentially, such as an EGD followed by PTG, including preoperative planning. At times, either treatment, surveillance, or surgery was warranted or required postoperatively, such as PTG dilatation of the GI tract, dumping syndrome, or relief from an obstruction.

One factor shared by some participants at this stage involved needing to make a rapid decision about choosing an intervention to undergo PTG after hearing about HDGC from a family member, when witnessing a family member diagnosed with HDGC, or knowing persons having surgery to rapidly remove the risk for cancer. For example, nine participants, who had witnessed family members dying from stomach or breast cancer or witnessed another with the CDH1 gene, immediately elected PTG surgery to avoid cancer rather than running any further risk by delaying this intervention. Two participants also considered prophylactic double mastectomy soon after the PTG because of a strong history of familial BC.

Age affected the speed by which participants decided to undergo surgery, as CDH1-positive participants older than 70 and younger than 25 were more likely to delay interventions.

Older participants tended to wait to have surgery and would take more time to seek the opinion of HCPs and genetic counselors to determine whether surgery was warranted due to age and other existing comorbidities. Similarly, younger persons were inclined to delay interventions as they considered family planning age and family history of BC as factors for BC surveillance vs. prophylactic mastectomy and multiple endoscopic biopsies prior to PTG. For those planning or already raising a family during their middle 20s to early 40s, conditions that affected the timing of interventions included family planning, career considerations, practical issues like health and insurance benefits, and managing family household duties.

Resources that helped the participant make intervention decisions included physician consultation, genetic counselor recommendations, peer-to-peer online sharing, communication with reliable bloggers, and data available at nonprofit support sites such as NSFC. Self-advocacy was used to obtain referrals and recommendations about where and with whom to have the PTG, including surgeon expertise in PTG intervention. A majority of the CDH1-positive participants expressed relief in obtaining access (n = 13) to travel away from home for the PTG based on the physician's level of expertise. One participant shared "Because a lot of the times that's what happens when the people have some kind of a problem elsewhere where they had the surgery,—I just would rather go to the experts." Further ability to access expert surgeons was also influenced by financial considerations, such as insurance coverage.

The iterative nature of the decision process in Stage 3 would lead to Stage 4—*normalizing*. After adjusting in Stage 4, the participant is often led back to Stage 3 where the next intervention decisions are made, leading to ongoing adjusting and additional treatment on an individual level of need (see Table 2; Stages 3 and 4).

Managing the initial intervention, subsequent treatment, or additional surveillance described the work of participants during Stage 4. This stage harkened the beginning of *normalizing* through adjusting to the changes brought on from the interventions chosen in Stage 3. Adjustments at Stage 4 included managing post-PTG motility modifications, surgical complications, dietary changes, body image adjustments, fatigue, and planning for generational concerns in the family, including future testing and surgery for the children and future family planning.

Stage 4

Stage 4 involves *normalizing,* including adjusting postoperatively. The iterative nature of Stage 3 and Stage 4 are related in part to the level of complexity especially when a cancer diagnosis was present prior to a person learning they were CDH1-positive (see Figure 1). Interventions were completed while new interventions would also be chosen, such as surveillance, surgery, or further treatment. Timing varied in both speed and order, such as how and when to consider surveillance for BC after completing gastric surgery with PTG, or vice versa in surveillance with endoscopy after undergoing a mastectomy. Such further interventions included PTG, double prophylactic mastectomy, endoscopic biopsies, and BC surveillance, including alternating MRI, ultrasound, mammography, and chemotherapy. Some participants reduced the layers of decision-making such as electing the PTG without surveillance biopsies. Other participants needed other interventions beyond monitoring the PTG adaptations, such as BC surveillance or deciding when to choose a prophylactic double mastectomy. After each intervention was chosen and completed, priority was given to adapting through follow-up and adjusting by monitoring. Further discussions about the timing and choice of additional surgical interventions or surveillance and treatment, if required, took place as *normalizing* occurred, all

leading back to Stage 3. A participant who was 6 months post-PTG was considering where to have her double prophylactic mastectomy and noted this: “So (not being alone again) was a big deciding factor with changing the location and having it (mastectomy) done here at my hometown at one of the local universities.”

Physical changes and gastric complications were reported after PTG (see Figure 4). Weight loss was a common response. One participant noted, “I'm struggling to put weight back on, but that's also fairly normal.” Two participants spoke of looking in the mirror with sadness regarding skin turgor or muscle mass changes. Social challenges were reported most especially in public situations where others might watch how much or what they eat, comments about their change in body type, and having their PTG compared to bariatric surgery for obesity. Psychological adjustments ranged from “not a big deal” and “I feel so tired” to “I just want to feel like myself again.”

Two participants reported a few adjustments to the PTG but were more concerned and dreading the next set of decisions regarding breast health and cancer risk. Emotional support was critical throughout Stage 4, including HCP listening, peer-to-peer support on the Internet through social media, and personal counseling sought by a few. One participant, when asked about any advice during the adaptation phase of her surgery, shared her approach as “It's really more of a mental game than the actual physical because the physical is something you can't really change.” Physical comparison before and after PTG was noted by one participant:

> I couldn't even physically open the hospital door because the door seemed heavy. And I'm just not used to… like normally I’d be “Hey I got that!” I would be strong; I'd be the first person to have moved something.

Another participant also indicated, “One of the things I have learned is that I have to be a really good advocate for myself.”

Self-advocacy after PTG included informing HCPs about which oral medications were tolerable or malabsorbed without a stomach, such as iron or B12. An older participant described the new rebalancing after PTG: “My iron was running a tiny bit low. The B12 was running low. The D was running a little low. I take supplements for all of those.... They’re now all within the normal range.” The length of the adjustment period varied for participants to adopt a new diet, smaller more frequent meals, and social situations involving intolerance to food.

Resources for learning what was expected pre- and post-procedure were identified consistently within distant specialty centers, sometimes locally, and often through peer-to-peer online support. As in Stage 3, participants sought others who had similar issues or experience. Participants were often managing multiple issues during Stage 4, such as caring for a family while undergoing treatment, seeking a nutritionist or registered dietician, family-planning considerations. They solved their challenges in this stage by reaching out for practical health and psychological support with peer-to-peer social media, including internet bloggers, while others sought traditional support groups and attended meetings, such as the NSFC Spotlight events. Overall, the four stages revealed that *Paving the way* is a complex process that accounts for individual conditions, contexts, factors, and influences leading to living longer.

Implications for Nursing

This GT study adds to the body of nursing knowledge through describing a substantive theory informing decision-making at each stage for those diagnosed with the CDH1 marker. The substantive theory provides nurses a roadmap for the patient diagnosed with the CDH1 marker to understand and assess the factors, influences and conditions at every stage . This GT model can

be utilized in further testing of influences and conditions as interventions to ameliorate anxiety and fears while inform patients better throughout the different stages of decision-making for the patient and the family after the diagnosis of the CDH1 marker. Nurses may assess for self/family advocacy, shared decision-making needs, resources such as peer to peer support, and navigating through life-event concerns within the complexity of care when cancer is found first for those diagnosed with the CDH1 marker. The development of a postoperative physical adjustment scale would also help predict further physical and emotional interventions such as managing nutrition and genetic risk in pregnancy and family planning. The study of influences of peer-to-peer social media influence is also warranted.

Nursing assessment should occur with these patients throughout the process of pre-and post-procedures including management of genetic testing, treatment, surveillance, and surgical interventions and post-surgical consequences. Patients living longer through preventative genetic decision-making need a nurse in the role of survivorship coach and throughout the adjustment process with wellness navigation. Evaluations post-operatively including appropriate follow-up services with specialized registered dieticians through nurse-navigation with the patient are essential for wellness post PTG.

Knowledge Translation

The findings of this study demonstratethat patients with the CDH1 marker face a complex process of problem-solving throughout this decision-making process including influences and conditions that occur from the entry point while faced throughout lifetime. The influences and conditions found in this study of age, advocacy, and the timing of life events compare to the following studies: (a) *Life Course Perspective* (LCP) grounded theory of *Living with genetic vulnerability* (Hamilton, Innella, & Bounds, 2016a,2016b); (b) a case study of

pregnancy after PTG (Kaurah, Fitzgerald, Dwerryhouse, & Huntsman, 2010); (c) the predictive genetic testing grounded theory of *Engagement* (McAllister, M., 2002); and (d) the recent virtual model program in oncology for navigation to promote self-advocacy (Schaffer, Haag, Borazanci, & Von Hoff, 2019). In the LCP, age was the primary factor of focus subdividing the cohort into groups—*timing of events* (20s), *human agency* (30s), and *linked lives* (40–50s)—all of which are comparable to the factors of age, advocacy, and life events throughout the stages of this study.

The findings of this study also reflect the need for persons with the CDH1 marker to make decisions to avoid or detect cancer early to preserve their health and have longer survival. *Paving the way* is comparable to other grounded theory research on the BRCA gene and ovarian cancer. Three grounded theory studies concluded that the goal of decision-making for patients with the BRCA syndrome included *Preserving oneself* (Howard, Balneaves, Bottorff, & Rodney, 2011) and *Maximizing survival* (Jeffers, Morrison, McCaughan, & Fitzsimons, 2014), and, with patients experiencing ovarian cancer, the goal of facing a life-threatening cancer is *Preserving oneself in the face of uncertainty* (Pozzar & Berry, 2019).

Several decisions were made throughout the process of *Paving the way* from the time participants were first made aware of their risk of having a genetic syndrome involving testing, undergoing procedures and/or surgery. A qualitative descriptive study of "*previvors*" for hereditary breast and ovarian cancer needed resources in four stages: pretesting, posttesting, premanagement, and postmanagement (Dean et al., 2017). A Norwegian grounded theory described that participants handled emotions while waiting for a gastric diagnosis by *Preparative waiting* that outlined a balance between "hope and despair" (Giske & Gjengedal, 2007, p. 90). An Australian registration study reviewing predictive testing for hereditary colorectal cancer syndromes confirmed that participants declined testing for numerous reasons involving trusting

the resources, benefits outweighing risks, and a need for witnessing positive outcomes over negative before electing testing (Keogh, Niven, Rutstein, Flander, Gaff, & Jenkins, 2017). This study revealed that most, but not all participants decided to share information that family testing was recommended. They deliberated on the timing of when and how to tell due to age, life events, and to whom to share with in the family, which is consistent with patterns of disclosure to families at risk for Huntington Disease (Klitzman, et al., 2007). This study's finding of interventions and adjusting to life through *normalizing* compares to the grounded theory studies of patient survivorship after diagnosis of Hodgkin lymphoma—*Transitioning to survivorship* (Matheson et al., 2016)—and the BC survivor—*Reclaiming life on one's own terms* (Sherman, Rosedale, & Haber, 2012)- the person adjusting to a changed life with the diagnosis of multiple sclerosis (Satinovic, 2017) and gaining normalcy as one re-engaged and re-invigorated mentally and physically after the disruption of a cancer diagnosis (Walker, R., 2015).

Conclusion

The findings from this study suggest that *Paving the way* offers nursing greater understanding of the process for the person diagnosed with the CDH1 mutation. Applying the model will aid in navigating the layers of complexity in their decision-making.. Nurses are in a unique place to serve as genetic nursing counselors in high-risk oncology clinics, pre- and postsurgical intervention as navigators, educators, and advocates for those diagnosed with this rare genetic syndrome and their families. The shortage of certified genetic counselors, especially in rural areas, positions nursing to further develop the specialty role of the genetic-counseling nurse to assist the person diagnosed with rare markers like CDH1.

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Table 1. Participant Demographic and Clinical Characteristics (N = 20)

Demographic Data	**Participants**		**Family Members**		**Healthcare Providers**	
Gender	**Female** N = 10	**Male** N = 2	**Female** N = 1	**Male** N = 4	**Female** N = 2	**Male** N = 1
Age Range (years)	23–77	61–70	20–72		32–46	
Average Age (years)	50.7	65.5				
Race						
White/Caucasian	9	2	5		2	
Black/African American	0	0	0		0	
Asian	1	0	0		1	
Native Hawaiian or Pacific Islander	0	0	0		0	
American Indian/Native American	0	0	0		0	
Ethnicity						
Hispanic/Latino	1	0	0		0	
Hispanic/Non-Latino	9	2	5		0	
Marital Status						
Married	8	2	0		N/A	
Separated	0	0	0			
Single	0	0	3			
Divorced	1	0	1			
Other	1	0	1			

	CDH1 Participants & Family Members (N = 15)				
Generation diagnosed with CDH1	First N = 6	Second N = 5	Third N = 3		Fourth N = 1
Years since diagnosed with CDH1 gene	**Female** N = 10	**Male** N = 2	**Female** N = 1	**Male** N = 2	**Family members who did not test** N = 2
Range	<1 year–>9 years	<1 year–>4 years	1 year–>12 years		
Average	2.2 years	2 years			

Surveillance biopsy				
Yes	3	1	1	N/A
Procedure, average number	2	2	4	
Mapping biopsy only?	7	1	2	
Total Gastrectomy				N/A
Yes	10	2	3	
No	0	0	2	
Chemotherapy				
Yes	4	1	0	
No	7	1	0	
Breast Cancer diagnosis before CDH1 discovery?				
Yes	3	N/A	0	
No	7		1	
Screening for breast cancer risk?				
Yes	7	0		N/A
No	0	2	3	
How often screening completed?				
Every 6 months	7	0	0	N/A
Yearly	0	0	0	
Every 2 years	0	0	0	
Other	0	0	0	
Stomach cancer or in-situ cancer found after total gastrectomy?				
Yes	9	2	3	
No	1	0	0	

Table 2. Stages, Core Category, and Accompanying Participant Narratives

Paving the Way	**Decision-Making**	**Verbatim Quotations**
Stage 1		
Learning of Genetic Susceptibility (Pre-testing)	Choice is to share or to not share the news, listen or not listen to the news.	We all got together for a big family meeting to discuss that she had breast cancer and that they were trying to figure out why it had happened.
Learning of genetic susceptibility is discovered through one of three entry points.	Entry points of discovery of the news appear via 1. Cancer found first, 2. Family member sharing, 3. Healthcare provider referral.	My daughter had stomach cancer and they discovered it down in (X healthcare facility). Her diagnosis sheet was on her (other daughter's) kitchen counter, and my other daughter read it, and read it was hereditary diffuse gastric cancer. She said, hereditary? So, she looked it up and saw, gee is this something in the family? My mom received a phone call from a cousin from her dad's side saying that they carried a gene and that she likely had the disease (gastric cancer) because she (the cousin) ended up inheriting it. I didn't know that I was at risk. I asked to have the genetic testing due to my lobular breast cancer diagnosis. I was told I was not a candidate due to my family, no direct family history. I insisted upon having genetic tests, a genetic panel was done. I put this message out there (by social media to the family) because I want to keep them informed, to let them know if you do want to get tested, go get tested. Well, once I knew they assumed it was from my father's side. I suggested to my mother, father, sister that everyone get tested because it is a dominant gene and hereditary. Then

		I contacted my second cousin and she said, "Oh, I have that too." That means that it was on my mother's side of the family.
Stage 2		
Discerning Genetic Susceptibility (Testing) Knowing the risk and confirming it. The process involves believing the risk exists and acting upon this knowledge.	Choices are to test or to not test for the marker. Testing is immediate, delayed, or not done.	Since my mother had died and my two uncles had died from stomach cancer. We didn't know it was CDH1. So, then I went and talked to a genetic counselor on the phone. Well, my mother is dying of diffuse cancer, and her oncologist, who I thought was great, he had enough good sense to check her for this mutation. She found out. She told all of us….I did a little research on it…So I wasn't sure what to do, but I thought I've got two young boys and we had a 50-50 chance of getting it so I thought, well I got to get it done. I had to know. Just like, yeah, (it) sits in your head. I guess I felt a sense of urgency too. I think that I was complaining about ongoing heartburn and stomach issues and he (the doctor) had already known that I carried a risk, so he encouraged me to get tested. I put it (testing) off because for me it was more of an insurance issue because I wanted to make sure that my career and my life insurance and my disability insurance were in place and then I want to get a house and then have my mortgage and all that before I found out I may have a risk for cancer. He has several times spoken to his different physicians, and he still hasn't had any testing. But I know he thinks about it from time to time. And I don't really say anything unless he brings it up.

Stage 3		
Choosing Interventions Choosing cancer prevention and potentially lifesaving interventions through accurate information. Choices include treatment, surveillance, and/or surgery.	Choices are to treat a found cancer and/or avoid cancer. Resources are utilized. Individual circumstances considered. Interventions are chosen one at a time in a personalized order, immediate, or delayed, or not chosen. Stages 3 and 4 are iterative. Stage 3 is revisited and leads back to Stage 4 for each next decision in the process.	Proactively I can cure this if it hasn't spread, you know, metastasized. It was recommended that I get my entire stomach removed. There were a lot of questions. When is the best time to do that? How will it impact my life? How will it impact having children? Obviously, there's no convenient time to get your entire stomach removed. I have a job; my husband is still a student in school. My mother passed away from lobular breast cancer, and I'm probably going to get it (cancer) if I don't do this (mastectomy). It was the internet, literally internet.com. It's been a rough 10-week transition. I didn't know anything about cancer until it came upon me, I didn't know anything about CDH1 until it happened to me. But I certainly spent a lot of time researching and finding out what I can do to be protected, to help myself. For my double mastectomy because of logistics, I might elect to have the surgery somewhere else, because it might be too difficult for me to do follow-up care in (a far-off state). I still must sort that aspect out. He still needs to make his decision. We'll do the endoscopy;

		we'll see how that looks. We know, damn well, the endoscopy means nothing. I mean, there's cancer there, like me, I was stage III.
Stage 4		
Normalizing Interventions are completed and new interventions of treatment, surveillance or surgery are chosen. Physical, mental, and emotional coping after interventions and ongoing surveillance.	Postintervention choices. Surveillance, treatment, and/or additional surgery. Returning to Stage 3, the next decision is considered.	They came back and said that he did have the signet ring cancer cells, so he did have cancer. It was super-duper early; we didn't expect to find much other than that. I don't have a stomach and that causes havoc. You're starting over again with the foods. I plan my time and my energy carefully. I plan when I'm going to eat. You don't have hunger. I must make myself go eat lunch. It's not like suddenly, I'm starving. You don't have that feeling. Your esophagus seems to get smaller. I have trouble sometimes when I'm eating, especially at dinner time. It just doesn't want to go down. I used to get up and just walk around the room. What happens is you just get used to all of it. It's not any big deal anymore. The second round, after major surgery and no longer having a stomach, it was much more difficult. So, my biggest adjustment, still, is I'm too skinny. I'm struggling to put weight back on, but that's also normal. I'm sort of just riding out, and saying this too shall pass, and just trying to do everything I can to shove food into my face.

	They thought of our family history (of breast cancer), annual MRI starting at 25, and at 30, we're (mother and daughter) getting MRI mammograms every 6 months, then colonoscopies starting at 30.

Table 3. Reasons to delay or choose to not test for CDH1 vs. more rapid testing

	Delayed or No Testing	Rapid Testing
Clinical	No recommendation from HCP No GI symptoms HCP declined referral to test	HCP insistence on testing GI symptoms Gastric cancer found Breast cancer found
Practical	Life insurance purchase Home purchase College classes New career	Health insurance to end new job Aging out of participant for healthcare coverage
Psychological	Serious family illness resulting in death Conscious denial	Witnessing others very ill from CDH1

Table 4. Participant (N = 17) CDH1 Testing and Surgical Interventions

Participant #	CDH1 marker +/-/N/A	PTG Y/N/N/A	Breast Cancer First Y/N/N/A	Curative or Prophylactic Mastectomy
1	+	Y	N/A	N/A
2	+	Y	N	N
3	+	N	Y	Y
4	+	Y	N	N
5	+	Y	Y	Y
6	+	Y	N	N
7	+	Y	Y	N/A
8	+	Y	N	N
9	+	Y	N	Y
10	+	Y	N	N
11	+	Y	N	N
12	+	Y	Y	Y
13	+	N	N/A	N/A
14	+	Y	N	N
15	+	Y	N	N/A
16	N/A	N/A	N/A	N/A
17	N/A	N/A	N/A	N/A
Total = 17	CDH1 + = 15	PTG Y = 13	Breast = 3 GI SRC = 1	Total = 4

Note. CDH1 = Cadherin-1 gene; PTG = prophylactic total gastrectomy; N/A = not applicable; Y = yes; N = no; GI SRC = gastrointestinal signet ring cell.

(see figure 4 below)

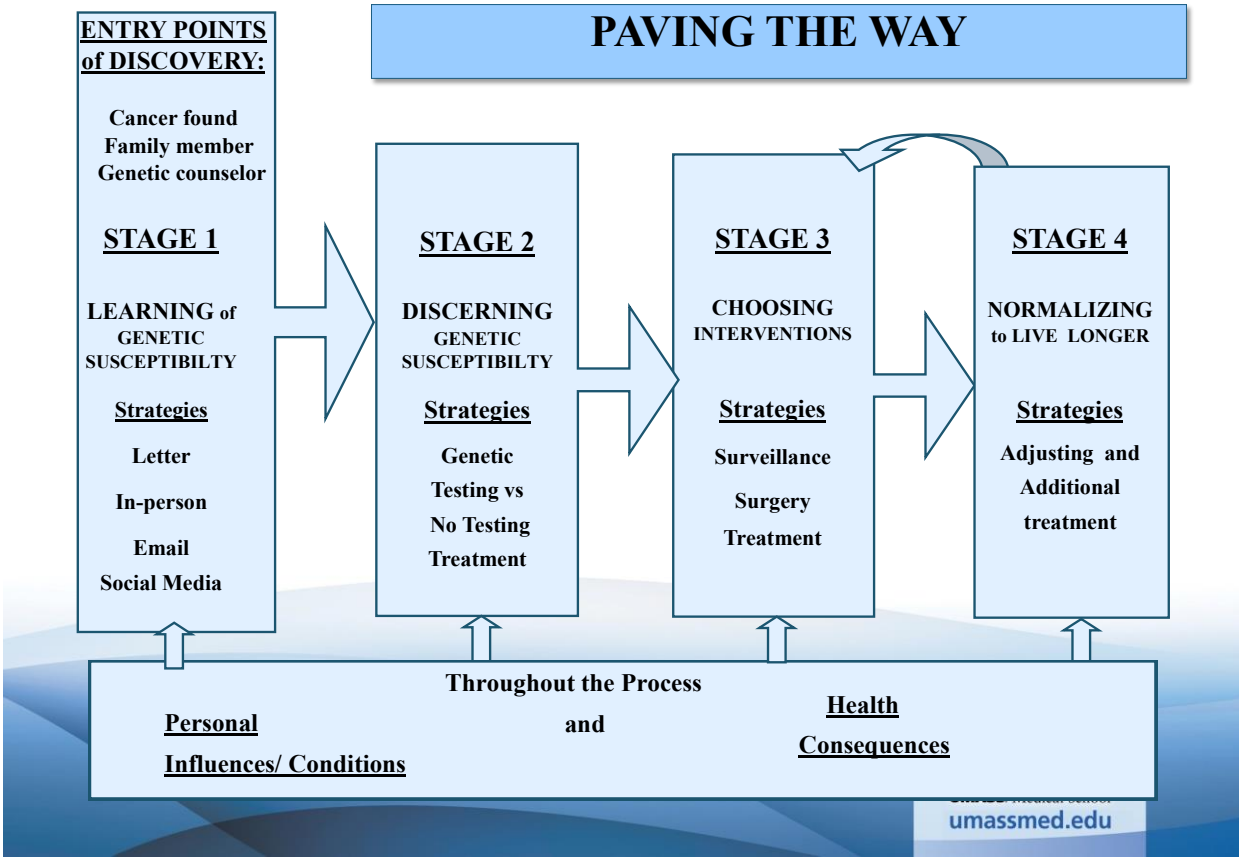

Figure 1. Paving the Way: A Grounded Theory.

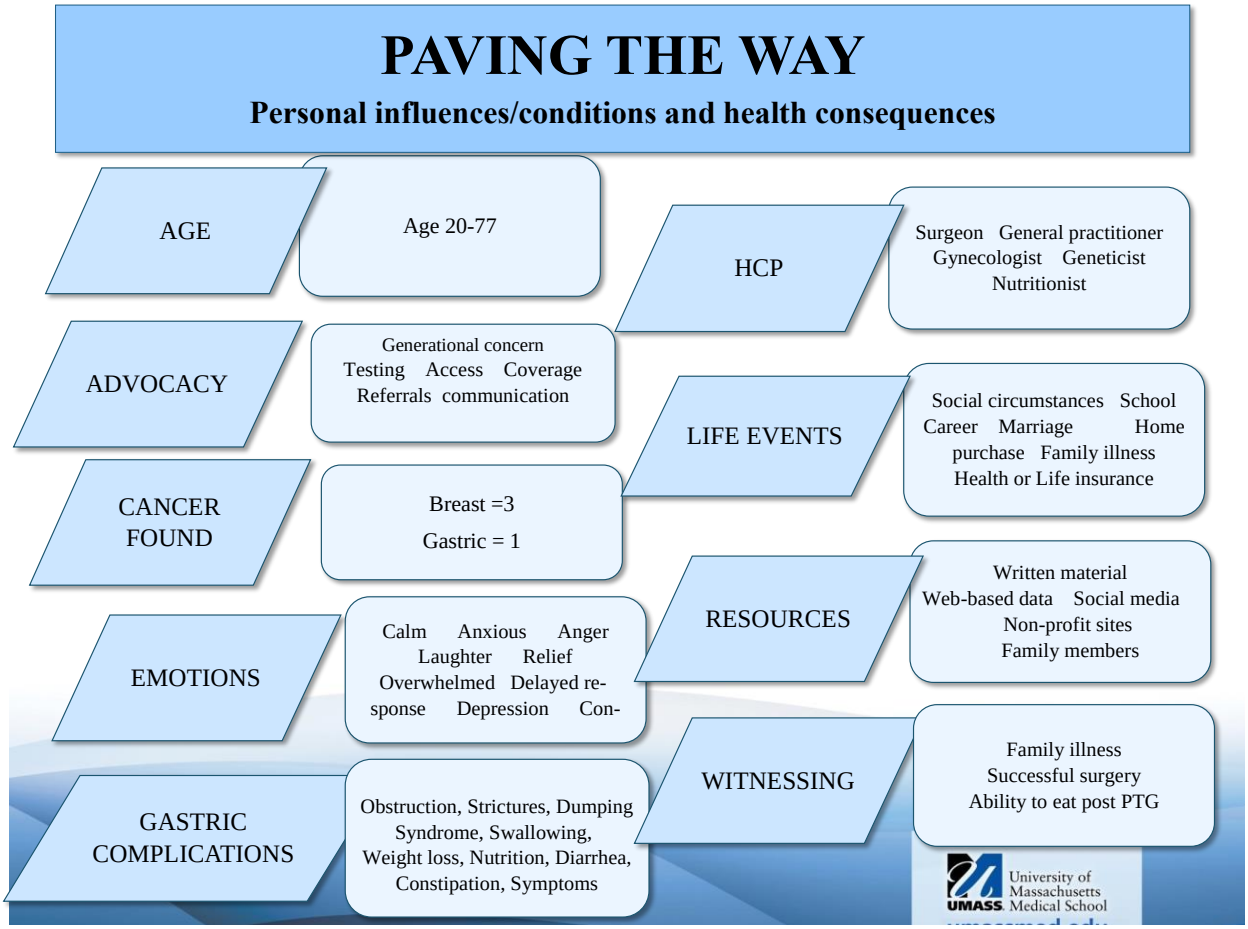

Figure 2. Influences, Conditions, and Consequences

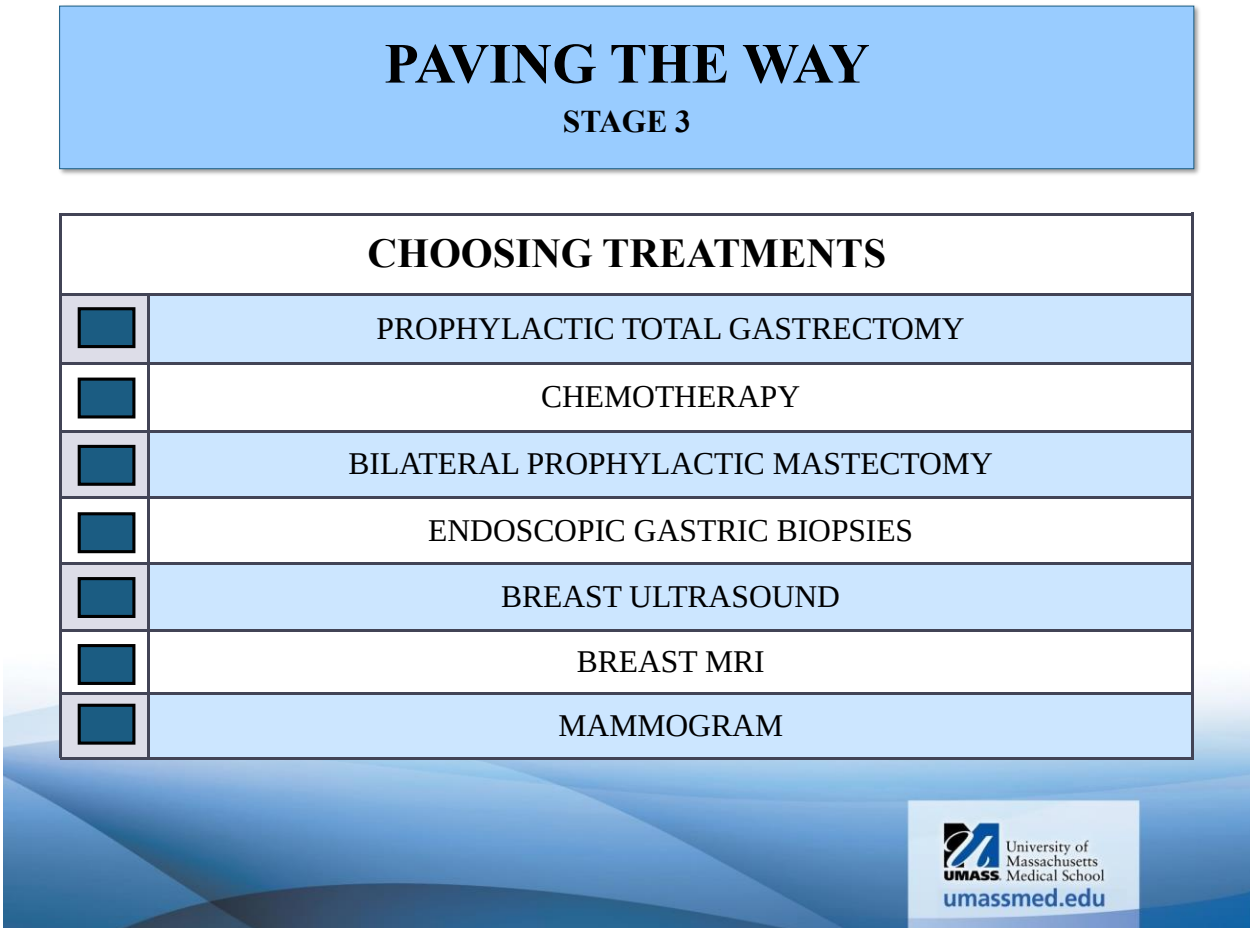

Figure 3. Choosing Interventions

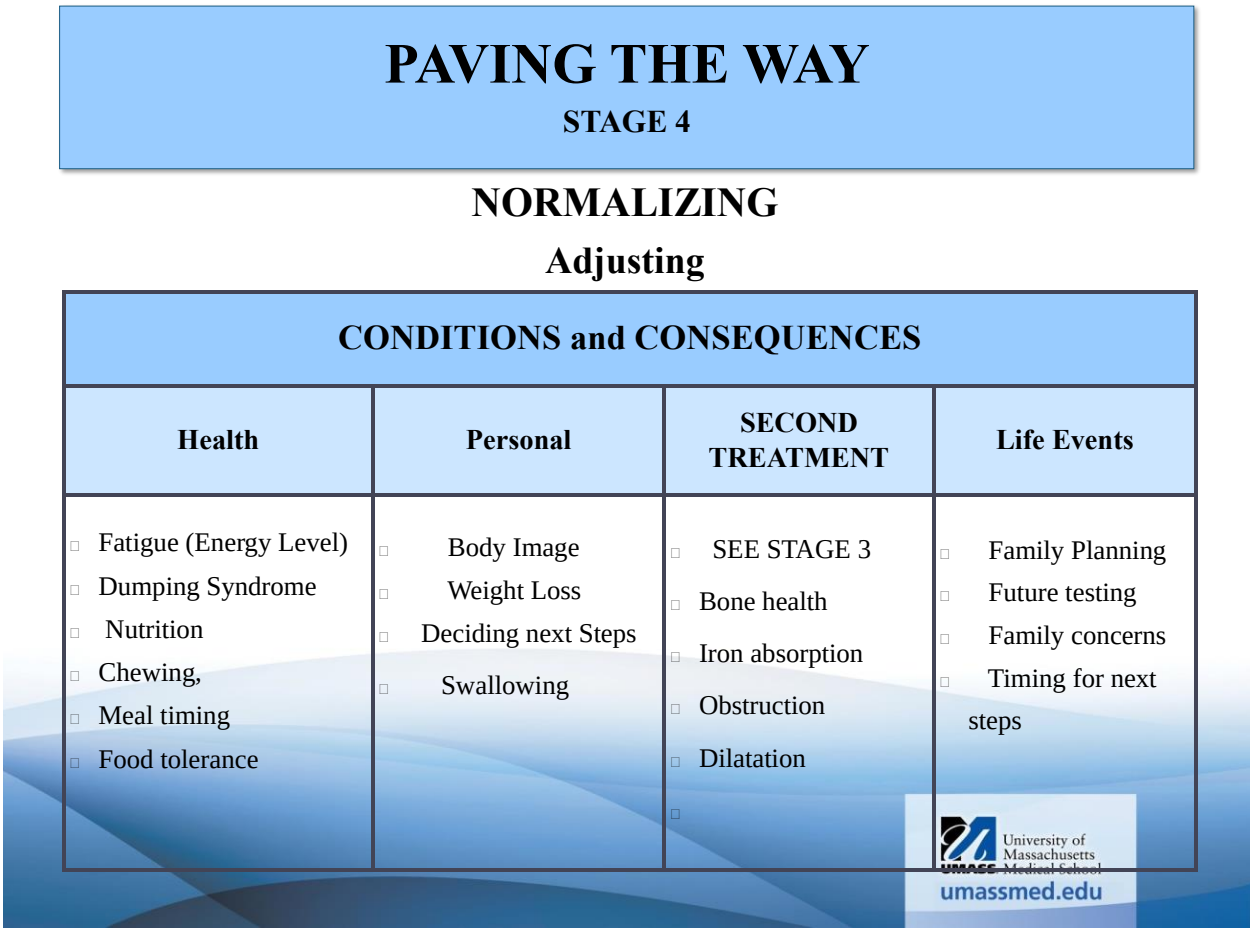

Figure 4. Normalizing and Adjusting