**Factors that Contribute to Errors in Risk Interpretation Based on Bayesian Analysis**

While determining an accurate risk figure using modifying factors may seem straightforward, cognitive shortcuts that lead to errors may alter interpretation of risk. Three common errors in Bayesian reasoning may lead to miscommunication or misinterpretation of risk (reviewed by Konheim-Kalkstein 2008):

- *Conservatism* is the process by which a person focuses only on prior probabilities and does not incorporated newer evidence into the risk assessment (Edwards 1968).
  Example: A patient has a family history of Marfan syndrome, an autosomal dominant disorder of connective tissue. The patient believes that all of the children she has will have Marfan syndrome. The patient neglects to take into consideration that the physical examination does not reveal any of the diagnostic criteria in her children.
- *Base Rate Neglect* is the opposite of conservatism and occurs when a patient only factors in newer evidence and does not consider prior risks (Kahneman and Tversky 1973).
  Example: A patient has a negative colonoscopy at the age of 25 and considers himself at decreased risk to develop colorectal cancer. He chooses to disregard his family history of colon cancer.
- *Inverse Fallacy* occurs when a person equates the hit rate (the sensitivity of the test) with the posterior probability. Of errors performed in Bayesian reasoning, the inverse fallacy is the most frequent (Villejoubert and Mandel 2002). A person who would exhibit pure inverse fallacy would estimate that having a child with Down syndrome following a positive NIPT is 99% rather than the posterior risk of 61% in the example above. The magnitude of this fallacy became more apparent when low risk women began having NIPT since the gap between the detection rate and their posterior risk is much wider.

Consider the following example of cognitive errors:

A two-day old male infant undergoes Newborn Metabolic Screening. The result indicates that he has a positive screen for galactosemia. The laboratory reports a 98% sensitivity of the test and a 0.05% false positive rate for galactosemia screening. The infant's a priori risk for galactosemia is 1 in 60 000 since there is no family history of galactosemia. In this example, only considering the infant's background risk (1 in 60 000) to have galactosemia is considered a conservatism error. Base rate neglect occurs when one does not consider the fact that the a priori risk is 1 out of 60 000 and only considers the screening information (the sensitivity of the screening). In such a situation, the parents might estimate that their infant's chance of having galactosemia at nearly 98%. If the parents fall into the trap of inverse fallacy, they are confusing the sensitivity of the test with the probability of having the disease given a positive result in the screening (they would conclude their newborn's chance of having galactosemia is 98%). They are neglecting the a priori rate (which is low) and the false positive rate. The actual posterior risk for the baby to have galactosemia using Bayes' formula is 3.2%. This example illustrates why it is helpful for the genetic counselor to understand the common errors that might occur in patients' understanding of risk assessments.

## Risk Communication

> Getting too deeply into statistics is like trying to quench a thirst with salty water. The angst of facing mortality has no remedy in probability. (Kalanithi 2016)

Communication is essential within the practice of genetic counseling – importantly it is not just conveying genetic information but also assessing patient comprehension (Henneman et al. 2008). This nuanced approach, utilized by genetic counselors, calls for assessment, analysis, and application of crucial genetic counseling skills to engage the patient in a thoughtful conversation in which risk is conveyed and the meaning and value of risk is explored to result in understanding by the patient (Green et al. 2004; Joseph et al. 2010). Genetic counselors who use this unique skill set as described in the Reciprocal Engagement Model (REM) are then able to guide the discussion to help anticipate preconceived concerns that a patient may have and gain a better understanding of their source (Veach et al. 2007).

## Strategies to Communicate Risk

Communicating risk information in a genetic counseling context is ideally an open, two-way exchange of information leading to better outcomes (Ahl et al. 1993). However, the specific communication strategy selected is typically based on the goal that the genetic counselor is trying to accomplish. For example, risk communication may look different if the goal is retention of information versus alteration of risk perception, influence on a health behavior, impact on anxiety or stress level, or dissemination of information within families. As opposed to websites and risk calculators, genetic counseling sessions afford an opportunity for genetic counselors to gauge the impact of their words on their patients and assess if the message is coming across in the way it was intended (O'Doherty and Suthers 2007). When determining which communication strategy to employ, genetic counselors should bear in mind that patients' post-encounter perception of risk will be influenced by both their preconceived risk perception and the risk information provided by the counselor.

### Communicating Quantitative Risks

Because quantitative risk communication is challenging, it is important for genetic counselors to consider the format they use to present risk numbers. A medical study by Gurmankin et al. (2004), reported that people preferred to receive risk information in a numerical format from their physicians; receiving numerical statistical information increased people's confidence and trust in the physician. Hallowell et al. (1997) likewise suggests that most people prefer that genetic counselors use quantitative data, supported by qualitative descriptions to communicate risks. Within the medical community, it is considered best practice to communicate risk in quantitative terms (Hallowell et al. 1997; Veach et al. 2018). However, many people have difficulty comprehending numerical information, particularly probabilities. Those with greater difficulties in comprehension of numbers trust information more when it is presented without numbers, suggesting individual preferences for presentation of risk information exist as a function of numerical ability (Gurmankin et al. 2004). In the United States, many adults do not possess high levels of numeracy (Center for Disease Control and Prevention 2019, Uhlmann et al. 2009). The 2012/2014 Program for the International Assessment of Adult Competencies found that only 10% of adults scored in the upper two levels of numeric understanding (PIAAC 2012/2014). Because most patients have difficulty understanding quantitative risk information, as described below, genetic counselors should be strategic about how to present quantitative risk.

- *Consider the Format in Which to Present Quantitative Data*: The same quantitative risk may be reported in different ways: as a frequency (1 in 200); as a percentage (one half of 1 % or 0.5%); as an odds ratio

(10 : 1); as odds against (10 to 1); or as population comparisons (your risk is 10 times greater than the general population), (Hallowell et al. 1997; reviewed by Konheim-Kalkstein 2008). In the medical literature, most agree that risk is best understood when communicated with natural frequency representation and pictorial representation (Cosmides and Tooby 1996; Gigerenzer and Hoffrage 1995; Hallowell et al. 1997). When comparing different risks, patients may find it helpful if the frequencies presented have the same denominator (Gates 2004). Many individuals focus on numerators only (e.g. 4 out of 100 or 2 out of 50); therefore, using consistent denominators can help (Reyna and Brainerd 2008). For example, instead of telling a patient who has a 1 in 100 risk to have a pregnancy with trisomy 18 on her screening test that the risk for miscarriage with amniocentesis is 1 in 500 and the risk for trisomy 18 is 1 in 100 or 1%, the counselor might state that the chance for trisomy 18 is 5 in 500 and the chance for miscarriage is 1 in 500. Utilizing the same denominator also helps to improve the understanding of the difference between numbers that are less than 1 (Okan et al. 2012). However, patients may also have a hard time visualizing 1000 or 100 000 people in a room, which means that smaller denominators can be more effective (Garcia-Retamero and Galesic 2009, 2011). Overall, providers should aim to eliminate or minimize patients' need to spend energy on mental calculations.

- *Present Information from Multiple Angles*: Consider the following: "Michelle has a 60% risk of developing breast cancer" and "Michelle has a 40% chance of *not* developing breast cancer." These are mathematically equal statements, but they are not psychologically equivalent. It is beneficial to attend to the way patients' frame the information themselves, while remembering that it is not possible to "assess the objectivity of [patients'] risk perception based solely on their ability to cite a correct probability" (O'Doherty and Suthers 2007).
- *Use Qualitative Descriptors Cautiously:* Studies in the genetic counseling literature demonstrate that patients may use qualitative risk descriptions to arrive at an understanding of their posterior or modified risk (Bottorff et al. 1998; Hallowell et al. 1997; Sagi 1998). Many believe that risk information presented using qualitative terms or probability is more persuasive (Hallowell et al. 1997; Koehler 2001). Caution is necessary when providing risk information qualitatively because of the subjectivity with which it may be interpreted.
- *Mix Quantitative and Qualitative Presentations for Optimized Understanding*: Studies show that people prefer to hear risk information in either quantitative or mixed (quantitative and qualitative) formats (Hallowell et al. 1997). Pay special heed to literacy and numeracy. Using a variety of formats (e.g. numerical, verbal, pictorial) can improve comprehension, but genetic counselors should select their formats carefully based on the background and context of each patient. For example, graphics have been shown to improve understanding of risk, especially in those with lower education and numeracy (Lautenbach et al. 2013).

  Since people differ in their preference of format, the counselor cannot assume that any single technique is best. Assessing patients' use of verbal qualifiers may be useful in gauging how they perceive their risk. However, it is important to bear in mind that patients may use the same qualifier in different ways at different times.

There are many other strategies that can be employed to improve the risk communication process. First, providers should acknowledge that there are different, yet complementary goals at play. While the genetic counselor's task is to convey a set of complex and highly abstract factors associated with a specific risk, the patient's task is often to make significant health-related decisions about an event which may or

may not happen (O'Doherty and Suthers 2007). The genetic counselor uses risk to provide guidance, while the patient uses risk to make a decision. Thus, it is advantageous to contextualize the information as precisely as possible by elucidating the beliefs patients hold about the implications of the risk and how they might respond (Rothman and Kiviniemi 1999). To that end, it is typically recommended to avoid using relative risks, recognizing that it can be very challenging for some patients to translate population frequencies into personal risk. Instead, it can be helpful to provide absolute probabilities, specifying the period over which the absolute risk applies. For example, telling Mark, who was recently found to have a pathogenic BRCA2 mutation, that his lifetime risk of breast cancer is 7% is more helpful than saying that his risk has increased 58-fold. Furthermore, patients should be encouraged to use risk estimates to assist in decision-making and *not* as a predictor of future events (O'Doherty and Suthers 2007).

An awareness of and appreciation for different learning styles and teaching styles also can improve risk communication. For example, if a patient's learning style is known (e.g. verbal–linguistic, visual–spatial, kinesthetic, intra- or inter-personal), then the genetic counselor can aim to deliver the risk information accordingly (Boud and Griffin 1987). For the large portion of the population (65–85%) that can be classified as visual learners, visual aids can be used with varying degrees of effectiveness to illuminate magnitude of risk, relative risk, cumulative risk, interactions between risk factors, duration of risks, consequences, reactions to risk, and the uncertainty of risk (Lipkus and Hollands 1999). Regardless of a patient's learning style, it is imperative to:

a) provide the risk information in context (i.e. explicitly state how the new information fits into the bigger picture and how it is relevant to the patient);
b) use thoughtful and accessible language (i.e. avoid jargon and value-laden words);
c) be careful not to sound more definitive than the data supports; and
d) not over-value scientific information (Melas et al. 2012).

Similarly, genetic counselors should compare the benefits and limitations of using the following approaches:

a) lecture-based teaching models where they position themselves as experts and expect the patients to listen
b) a narrative, life history model where they engage in active listening to empower the patient to tell their stories; and
c) mutual participation models, such as the reciprocal engagement model of genetic counseling practice where they and the patient engage in a mutually interdependent investigation of the risk (Uhlmann et al. 2009; Veach et al. 2007).

A provider should also consider the service delivery model in use: Is the risk communication occurring in person or by phone/video? Is a decisional aid being used? Is the risk being provided individually or in a group? (Hilgart et al. 2012; Senay and Kaphingst 2009).

Considering the stage of life cycle development of the patient also is important when communicating risk. For example, when speaking with children and adolescents, genetic counselors should remember that youth tend to be accepting of new information, deem others as being responsible for their learning, and recognize that the newfound data may be mandatory for their future goals and/or subject to postponed application (Knowles 1968; Knowles et al. 2012). In addition, adolescents and young adults often have limited exposure to complex decision-making and are experiencing a variety of physical, social,

cognitive, and emotional changes, all while moving from concrete to more abstract thoughts (Piaget 1964). Taken together, these factors influence their perceived susceptibility to risk and thus their receptiveness to risk communication (Nikiforidou et al. 2012). Meanwhile, adults may seek out new information in a self-directed, yet skeptical manner, in the hope of accomplishing a specific goal (Knowles et al. 2012).

Adult Learning Theory (Knowles 1973) suggests that successful learning requires four key elements: motivation, reinforcement, transference, and retention. Therefore, if a patient does not recognize the need for the information, does not have the message reinforced, and/or is not able to use the information in a new setting, then the information will not be remembered, and the genetic counselor's effort will be in vain (see Chapter 13).

Family Theory may also be useful for determining how to incorporate risk information into the patient's life role, responsibilities, and family experiences (Bowen 1978). Consistent with Family Systems Genetic Illness Model, risk may be perceived differently during periods of crisis, transition, stability, adaptation, and resilience (Rolland 2006; Rolland and Williams 2005). Furthermore, genetic counselors' risk communication strategies should account for how patients compartmentalize risks to themselves versus risks to their children, grandchildren, siblings, etc. (Daly 2015; Gaff et al. 2007; Gilbar 2007; Rolland and Williams 2005). Genetic counselors wishing to learn more about factors that impact risk perception and communication should explore other theoretical models in addition to Adult Learning Theory and Family Theory, such as Theory of Planned Behavior (Ajzen 1985), Cognitive Behavioral Theory (Kendall and Hollon 2013), and Positive Psychology (Seligman and Csikszentmihalyi 2000) (see Chapter 9 for more on Family Theory).

## Influences on Risk Perception

Most patients have a defined idea of their genetic risk, although in practice, it is probably not quantifiable, or a person's idea or understanding of the true numeric value may be skewed based on personal experience (Austin 2010; Sivell et al. 2008). Individuals contextualize all risk so that the daily exposure to or acceptance of an event may increase or decrease the significance to the risk. Patients' context may include the etiology of the condition, its prevalence, their family history, their cognitive and emotional traits, their physical resemblance to relatives with the condition, lifestyle factors (e.g. occupation and diet) and other environmental factors (Ekwo et al. 1985; Sivell et al. 2008). Meanwhile, their previous perception of the nature of the potential outcome (i.e. the severity) may be colored by how a relative experienced the condition at hand, how closely they observed the progression of the condition, and their confidence in current or future treatment and management options (d'Agincourt-Canning 2005). Preconceived risks may also stem from interactions with family members, friends, physicians, or neighbors; or from books, magazines, or the media. A discussion of how preconceived risk impacts risk communication in the form of heuristics is discussed later in this chapter.

Although the patient's estimation of preconceived risk may be inaccurate, it typically continues to factor into the patient's perception of overall risk (Pearn 1973; Shiloh 2006). An informed patient in the genetic counseling setting is one who can make a decision based on preconceived risk ideas (subjective and objective), background information, and those risks discussed by a genetic counselor and explored within the context of what the patient brings to the session. In other words, perceived risk is a combination of a patient's context, the perceived severity of the condition at hand, and the numeric probability

that is introduced. Studies have shown that perceived risk is a stronger motivator for uptake of protective behaviors and preventive measures than absolute risk (Austin 2010; Sivell et al. 2008).

During the last several decades, a significant shift in medical care philosophy from a paternalistic viewpoint to one of team goals and family centered care has impacted risk communication as well. It is with this new expectation of collaborative decision making that many patients arrive to genetic counseling prepared with questions based on review of online sources and input from their support network (Edwards et al. 2008). Genetic counselors must assess and adjust their practice to meet the literacy and technology needs of their patients. As communication in genetic counseling practice continues to incorporate, and in some ways become more dependent upon technology, it is vital to recognize that each patient has a different relationship with technology as a tool (Joseph et al. 2010). Educational tools may serve to help with overall patient understanding, but genetic counselors must not rely solely on them and lose sight of the value of the risk communication discussion to assess patient understanding and the personal variables that influence risk comprehension (Joseph et al. 2010).

Patient perspective is shaped by numerous lifetime factors. Further differences that complicate risk communication include ethnic and cultural backgrounds, generational differences, education, and direct experience. Furthermore, understanding of genetics, genetic testing, and the implications for disease management is impacted by gender, age, race, education level, cultural, immigrant and socioeconomic status. Barriers to risk understanding may be exacerbated by a lack of culturally-based materials on health and screening (Peters et al. 2007). The availability of culturally relevant information can help patients understand and accept risk. What the genetic counselor may initially perceive as a lack of understanding of risk may actually represent long standing cultural beliefs. It is, therefore, the responsibility of the genetic counselor to recognize the beliefs of their patients and engage them in a culturally relevant exploration of risk. Attempting to immediately correct preconceived ideas without an understanding of the patient's cultural perspective may undermine the risk communication (Peters et al. 2004). Additionally, genetic counselors must pay special attention to their own internal understanding of culture and how they communicate risk to different ethnic communities (Helms 1990).

Cohen et al. (1998) state various ethnic groups have different perceptions of risk as etiologies of birth defects. For example, in some cultures, behaviors during pregnancy are assigned risk that is perceived to have a large genetic burden. The use of scissors is prohibited during pregnancy in some cultures as it is thought to cause the fetus to be born with a cleft lip. Some people of Hispanic background may be less likely to consider medication exposure during pregnancy as a significant risk for fetal birth defects (Cohen et al. 1998). Alternatively, people from some religious and cultural backgrounds perceive birth defects as a punishment by God (Cohen et al. 1998). Additionally, cultural framing of risk contextualizes how patients hear risk. For example, in India, most women approach pregnancy with a fatalistic view. Many believe that a woman has no control over a pregnancy or the subsequent outcome of the pregnancy (Choudry 1997).

As another example, in some settings, women are expected to defer to the male partner. In doing so, their perception of risk may be modified based on their cultural norms of gender roles. Though specific cultural beliefs may dominate in a community, genetic counselors need to guard against assuming that all members of the community hold a similar belief without considering the particular personal experiences of each patient.

In addition to general cultural and social beliefs, genetic counselors should be mindful of family specific situations that influence risk perception. Family history plays a significant role in risk perception. A woman whose first three children have Phenylketonuria (PKU, an autosomal recessive inborn error of

phenylalanine metabolism) may be unwilling to accept that there is a 75% chance that her next pregnancy will not have PKU. Instead, she may focus on past history to modify her perceived risk (Bottorff et al. 1998). Alternatively, a young man who seeks genetic counseling because his father has Lynch syndrome (an autosomal dominant cancer predisposition syndrome) believes that because he favors his father's family, he has inherited the predisposition to Lynch syndrome as well. This fatalistic belief is the risk he is anchored to prior to the session (Bottorff et al. 1998; d'Agincourt-Canning 2005). The genetic counselor must work with the patient to identify this underlying belief and then discuss how the autosomal dominant inheritance of the predisposing pathogenic variant is independent of the genes for appearance.

Gender may also factor into perceived risk. In some cultures, male offspring are valued differently than female offspring.

| | |
|---|---|
| GC | (To a patient with a family history of Hunter syndrome) "I understand that you have a family history of Hunter syndrome." |
| PATIENT | "Yes, my brother and cousin both had Hunter." |
| GC | "How has this impacted your family? |
| PATIENT | "It is really tough. No males have made it in two generations." |
| GC | "How is that for your family?" |
| PATIENT | "It is very difficult. My mother desperately wanted sons, but she was afraid to try again. Now, she tells me that it is my duty to have healthy boys for her." |
| GC | "What is that like for you?" |
| PATIENT | "I am really scared. In my family, the goal is to have a healthy son. It is a lot of pressure." |

While understanding the context that the patient brings to the interaction is an essential element when providing risk estimates, another key element is the risk figure itself.

## Heuristics and Risk Perception

Heuristics play a large role in risk communication. From "eureka" which means "to find," heuristics are mental shortcuts for arriving at quick conclusions or solutions. These rules of thumb use an intuitive process to help us make sense of the world. In the context of risk communication, they influence how people understand numbers and risk and how they make decisions based on those numbers (Edwards and Elwyn 2001; Gigerenzer and Edwards 2003; Wertz et al. 1986). In moments of information overload and/or heightened emotional states, heuristics allow us to avoid considering everything at once, thus saving cognitive effort. Heuristics are usually effective; however, they can also lead to predictable and consistent cognitive errors. Common heuristics include affect, effort, peak-end rule, contagion, anchoring and adjustment, availability, and representativeness (see Table 4.8). Other related biases include gambler's fallacy, the Hawthorne effect, bracing, hindsight, confirmation, familiarity, bandwagon effect, and extreme aversion (see Table 4.9). Lastly, there are a variety of social biases that influence the impact of risk communication. For example, people tend to:

a) be overconfident in their own predictions;
b) think that they have control over events in general (and more control over events that impact themselves than others (fundamental attribution error);
c) accept more responsibility for successes than failures; and
d) believe that bad things are more likely to happen to other people (optimism bias) (Breakwell 2000; Klein and Helweg-Larsen 2002).

**Table 4.8** Common heuristics.

| Heuristic | Description | Example in Genetic Counseling |
|---|---|---|
| Affect | Tendency to rely on good or bad feelings | "I have a good feeling a about this doctor/hospital. I just don't think the cancer will come back." |
| Effort | Tendency to determine the quality or worth of an outcome from the perceived amount of effort that went into producing it | "We worked so hard for this pregnancy. I'm not willing to risk a miscarriage." |
| Peak-End Rule | Tendency to judge an experience largely based on how one felt at its peak (i.e. its most intense point) and at its end, rather than based on the total sum or average | "I was miserable during the first trimester and during labor. This pregnancy was a lot harder than my other ones." |
| Contagion | Tendency to avoid contact or associate risk with people or objects viewed as "contaminated by something bad" (or, less often, to seek contact with people or objects considered good) | "Both of my neighbor's kids have SMA. Our chance must be high." |
| Anchoring and Adjustment | Tendency to start with an implicitly suggested reference point (the "anchor") and adjust until a plausible estimate is reached | "I was told that the chance of a problem was really low based on my age. I don't think it could really be that much higher now just based on the ultrasound." |
| Availability | Tendency to rely on immediate examples that come to mind when evaluating a specific object | "I've never heard of Prader-Willi, and we don't have anyone in our family with it. I don't think that could be what Dylan has." |
| Representativeness | Tendency to assume that if a known object is similar to an unknown object in one aspect then it will also be similar in other aspects. | Patient: "My siblings are just like my uncle with hypertrophic cardiomyopathy. None of them exercise and they all drink and smoke, but I don't so I'm sure I don't have it."<br>GC: "All of my Latina patients are the same. They never want an amniocentesis, so I don't see the point of offering it." |

Genetic counselors should also acknowledge the impact of emotions on risk communication. Although using numbers gives the impression of a highly rationale process, emotional responses often take precedence over and alter factual information. Open conversation about emotions, cultural beliefs, and a priori perceived risks contributes to meaningful and effective discussion of risk (Bottorff et al. 1998). Genetic counselors can engage their patients in an investigation of their emotions, preconceived notions (and their sources), encouraging them to restate their gestalt/gist conclusions (i.e. the essential aspects or take-home messages) (Reyna and Adam 2003), and allowing ample time for processing and opportunity for questions. Taken together, these efforts can improve comprehension and adaptation by allowing patients to organize, categorize, and assimilate the information.

While the patient's a priori risk perception is integral to the outcome of risk communication, so is the value genetic counselors place on their own interpretation of risk, as this will influence the way it is conveyed (Henneman et al. 2008). Despite the best of intentions to maintain neutrality, there are a variety of subtle and overt messages that can be conveyed by tone, body language, order of and time spent on topics, and subjective modifiers such as high versus low, some versus most, and minimal versus significant. For example, those genetic counselors who place a high value on quality of life may present the benefits, risks, and limitations differently that those who place a high value on the longevity of life when discussing treatment and management options in a cancer setting.

Genetic counselors should also consider that the way risk information is communicated impacts patients' understanding and interpretation of risk. It is not just what is said, but also how it is said. As

**Table 4.9** Biases that may alter risk perception.

| Other Biases | Description | Example in Genetic Counseling |
|---|---|---|
| Gambler's Fallacy | The mistaken belief that, if something happens more frequently than normal during a given period, it will happen less frequently in the future (or vice versa) | "I have three unaffected children, so I'm sure the next one will be affected." |
| Hawthorne Effect | When people alter their behavior due to their awareness of being observed | "My kids are watching how I'll react, so I'm not going to show that I'm scared and nervous." |
| Bracing | To physically or mentally prepare oneself for something in an attempt to limit any adverse impact | "I always assumed I'd get Huntington disease, so I've been preparing for the worst all my life." |
| Hindsight Bias | Tendency to overestimate one's ability to have predicted an outcome that could not possibly have been predicted | "I knew it all along. This cancer spread too quickly to have been random." |
| Confirmation Bias | Tendency to interpret new evidence as confirmation of one's existing beliefs or theories | "I knew my headaches were a sign of something bad. Even though you say the MRI was normal, my increased fatigue tells me you must have missed something." |
| Bandwagon Effect | Tendency to do something primarily because other people are doing it, regardless of one's own beliefs, which may be ignored | "Everyone says your lab reports are confusing and impossible to interpret. I'm taking my raw data and to another company." |
| Extreme Aversion | Tendency to avoid choosing or believing in options that are at one extreme or the other | "You said the recurrence risk would decrease by 95% if I underwent surgery, but I bet it's more likely in the middle." |
| Familiarity Bias | Tendency to prefer or be more comfortable with familiar or well-known outcomes | "I grew up with kids who had cystic fibrosis, and their lives seemed fine. 22q deletion syndrome must be a lot worse." |

was discussed above, numerical presentations might include frequencies, percentages, odds ratios, and/or population comparison. Verbal presentations may include descriptors such as "high chance," "increased risk," or "positive." Visual presentations might rely on a patient's ability to interpret a jar of different colored marbles, a pictogram of stick figures, a pie chart, a line graph, or a histogram. Similarly, a patient's risk reception may be influenced by how many risks are presented (single vs. multiple) and the sequence of presentation in the case of multiple risks.

Furthermore, the way information is framed has a significant impact on the way it is received (Edwards et al. 1999, 2002; Edwards and Elwyn 2001; Gigerenzer and Edwards 2003; Lautenbach et al. 2013; O'Doherty and Suthers 2007). A patient who is told that there is a 1% chance that the baby will have intellectual disabilities based on the ultrasound findings (negative framing) will process the information much differently than if told that there is a 99% chance that the ultrasound finding is a normal variation that will lead to a healthy outcome (positive framing). In addition, it appears that the effects of framing vary depending on the situation. People tend to place a higher value on things they have compared to things they do not have (Tversky and Kahneman 1981). Similarly, people tend to feel more regret about actions that lead to a loss than losses associated with an inaction or missed opportunity. As a result, patients may prioritize avoiding losses over maximizing gains (Abramsky and Fletcher 2002; Melas et al. 2012; Tversky and Kahneman 1981). In addition, Shiloh and Sagi (1989) found that individuals are more likely to estimate a higher risk if they are asked to translate a verbal risk into a numerical risk. Welkenhuysen et al. (2001) concluded that individuals are more likely to elect testing if a verbal presentation is framed negatively, while they are less likely to be influenced by either positive or negative framing of numerical presentations.

## Summary

There are many factors that must be considered to understand how to best communicate risk, including the factors that influence how patients perceive risk, the value that genetic counselors bring to risk interpretation, and the strategies they use to explain and tailor risk. Though it may be difficult to achieve precise and accurate risk communication, it may be possible to achieve comprehensive risk communication that is sensitive to the patient's context (O'Doherty and Suthers 2007) and that respects the patient's request for guidance on how to manage and cope with their risk status (Sivell et al. 2008).

## Learning Activities

### Activity 4.1 Quantitative vs Qualitative Presentation of Risk

Break up into groups of two or three and each take a turn as the patient and as the genetic counselor (and observer, if working in triads). Use the following scenarios to role-play risk communication first using qualitative, and then quantitative terms: