

# Chapter 16

## Psychosocial Issues

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### Introduction

Upon learning the diagnosis of Fanconi anemia, the family must make important decisions that require a sophisticated understanding of a complex illness with many treatment options. While any serious illness in childhood can isolate a family, isolation is more likely with an unusual condition such as FA. There is often no inherent societal support for rare, unfamiliar diseases. The disease path for families affected by FA is interwoven with grief, loss, and uncertainty at every juncture. The diagnosis of FA imposes a change on the family system. The challenge is for parents to balance their ensuing sense of loss and grief, while orchestrating their child's medical care, maintaining hope, and sustaining a semblance of normal family life. FA is an illness with a course that is ever changing, allowing for much potential for optimism after the impact of the shock of the initial diagnosis.

### The Course of the Illness

FA presents different issues for families depending on the developmental stage of the child and the course of the disease for the individual child. Initially, before FA has been clearly diagnosed, families will be hopeful that the diagnosis will be incorrect. Without indisputable confirmation, there is always the hope that their child will not be severely affected.

The time of diagnosis itself is an emotional crisis; it takes time before parents can move from shock and disbelief to a more proactive mode of coping. The number of children with FA in the family, the number of unaffected siblings, and the ages of the children will affect the emotional profile and needs of a family at a given time. All families worry that they will not be able to learn enough about the disease to make good decisions for their children.

Depending on the age of the parents at the time of the diagnosis, the implications for the family are great. Will they have the physical or the emotional energy, the time, the desire or the financial resources to have more children? Will they arrange for prenatal diagnosis or histocompatibility (HLA) typing for subsequent pregnancies?

At any age, parents of children with FA often find themselves in the position of having to make difficult decisions, whether about medications or other treatment options. They may need help thinking through their choices and the implications of those choices. They need information that they can understand to make the best choice given the present state of knowledge.

Children with FA can be stable or asymptomatic for long periods of time. Emotionally calmer times may alternate with more volatile ones. As described in *The Damocles Syndrome*,<sup>1</sup> parents are constantly waiting for the next bad thing to happen. Helping families adjust to living each day to the fullest and to focus on activities apart from the illness are crucial components in day-to-day coping. The moments that are not driven by medical crises are times for families to learn and stay abreast of salient treatment options and to prepare themselves for the future. Living with uncertainty, and preparing

for a future with potentially complex medical situations, while helping a child embrace life and establish dreams, visions, and plans for the future, place the parent of a child with FA on a unique and challenging journey.

Should a child's condition deteriorate and alternate treatment options be considered, the family may be thrown into emotional crisis again and feel hopeless and immobilized. Being prepared to take appropriate action, feeling informed, and feeling supported, all help family members to move forward with the necessary tasks during these periods.

With some of the very difficult choices that parents will have to make for and with their children, there is no turning back. Therefore, each major decision requires that families and older patients know all they can prior to making the decision, with an opportunity to integrate the information and reflect upon and accept the choices they have made. In certain cases, families will be making decisions about experimental procedures and protocols which have been utilized with very few patients. Families experience a vulnerability and a unique anxiety when they know they are traveling on a road that few have traveled before.

## **Parents' Journeys**

Parents may cope separately and very differently with FA. One parent may need to learn everything there is to learn to plan strategically for the future, whereas the other may choose to stay focused in the moment. One parent may need to talk and to cry, whereas the other may not. Differences in coping styles as they relate to gender and culture should be recognized so each can be supported for his or her strengths, insight, and ability during the course of the illness.

If a marital relationship was previously stressed, difficulties in the relationship will often be exacerbated by the illness. On the other hand, some couples have felt that the strain and the magnitude of the issues they face have made them stronger together.

Depression and anxiety are two uncomfortable emotions characteristics that may accompany this disease. Many parents feel anxious or depressed from the onset, unsure of what to anticipate. The ability to contain the anxiety or depression, to make decisions, to enjoy life, and to continue to function are skills to be mastered.

Staying informed of current research and the ever-growing knowledge base about FA and potential treatments can help parents feel calmer, focused, and grounded. Talking to other parents, understanding their decision-making processes, and getting support help parents to maintain the balance they need. Counseling, information, and support from the Fanconi Anemia Research Fund, its e-groups, and communication with professionals play effective roles in helping with the ongoing adaptation of children with FA and their families. These support groups offer parents the opportunity to be parents: to be able to compare their child to other children, to seek companionship of another parent in a similar situation, to brainstorm, to share information, and to join the fight against Fanconi anemia and become empowered in the face of the illness.

Families may be viewed incorrectly as aggressive when they advocate in the interests of their children. There may be moments when families and individual physicians do not agree on treatment options and alternatives (e.g., hematopoietic stem cell transplantation). The involved professionals must work to make the best decisions with, and not for, families. This strategy will

help minimize potential later regrets for families and professional staff.

Relationships with their physicians are of tremendous value and significance to families affected by FA. The quality of these relationships often influences the family's entire experience of the disease. Helping navigate the course of the illness, and thinking through decisions can help those facing such rare illnesses feel much less isolated.

Families truly manage to become experts about FA. They must integrate tremendous amounts of information, while attending to their child's medical needs, and managing all the other activities of the family. It is not surprising that, when parents of children with FA are asked about what they've learned about themselves or their children since the diagnosis, they overwhelmingly suggest that they have learned how strong and capable they and their children are. Parents describe having a greater appreciation for the things they do with their children, learning how to experience each day to its fullest.

With ongoing innovations in technology and the refinement of preimplantation genetic diagnosis (PGD), some families are trying this option to facilitate having a child who could be a matched donor for a stem cell transplant for their child with FA. This process can be financially, emotionally, and physically draining and in some cases, all-consuming. Unsuccessful PGD attempts will serve to delay having more children and can create other conflicts for the family. This phase can be an emotional one in the life of an FA family, as treatment options as well as additional children stand in the balance. Successful PGD attempts, joyous in nature, can set the course of a family towards having a baby and planning a stem cell

transplant, creating an unusual dichotomy: anticipating the transplant and anticipating the birth of a child in the same instance. Families can benefit from talking with others who have been in this situation to help mitigate the intense emotions that can occur during this time. Parents who are of an advanced age and unable to utilize PGD as an option may experience remorse that this technology was not perfected earlier in the course of their child's illness.

### **Children with FA**

How parents accept and face the illness will influence how children with FA grow and adapt. If parents create an environment that allows for questions, discussions, and an expression of feelings, children will feel free to ask their parents about their illness and treatment options and become active participants in the disease management.

Children often know much more about what is happening than adults might believe. In addition to what they have been told, they pick up information from ambient conversation, have independent interactions with professionals, and surmise things from the emotional climate around them. They will ask questions when they want to know, and will often shy away from questions to which they do not want the answers. Children are good regulators of their own knowledge base, providing cues to the adults around them at all junctures.

Visible characteristics of the disease, such as the frequent short stature or missing thumbs of a patient, serve as a constant reminder to the outside world that the child with FA is different. At all ages, physical and other differences may set children with FA apart from their peers and can be factors which cause children

to feel isolated, lonely or depressed, affecting their self-esteem and ability to focus on age-appropriate achievements. Counseling can be a great benefit during these times. Children need to be able to confide in their parents and others when they feel limited physically or socially by Fanconi anemia.

A major concern of parents is what and how to tell children about FA. At each stage of development, children need age-appropriate explanations of their diagnosis and treatment. These explanations should grow in sophistication as the child grows. Information offered regularly to children will enhance their ability to understand their disease and establish trusting relationships. As they get older and medical problems emerge, groundwork set in earlier years will encourage patients to rely on health care providers.

School may present unique issues for children with FA. Some may have cognitive impairments that will require special attention. Others may have no known problems but, because of illness-related absence, may need extra assistance. School is the place where children may begin to feel as though they are unlike other children, if they are frequently absent because of doctor's visits, if they are sick and unable to attend, if they are unable to participate in activities or if they are perceived as different from their peers. They may need support to learn how to adapt, respond, and connect to their peers. Clearly assessing the child's educational and social needs, the educational program, and what works in a family will open discussion of these issues and allow for the best academic and social plan.

School-age children develop increasingly strong relationships with their peers as they begin to differentiate themselves from their families. Physical limitations

necessitating dependency may influence the child's social activities. Each child and family must find a balance in social and family relationships, which allows for a blend of independence and dependence, nurturing and differentiation.

Children with FA, facing multiple hospitalizations and medical treatments, are exposed to difficult experiences, including the deaths of other children or siblings. They may, therefore, come to understand and deal with issues with which adults may not feel comfortable. Although parents work to "normalize" their children's lives, patients' experiences are unique and force them to deal with issues associated with death at an "age-inappropriate" time, certainly at younger ages than other children. Thus, they may seem more mature than their chronological ages and often are more sophisticated than their peers in matters of illness and death. They may also appreciate life, and the meaning of life, more than the adults they encounter.

For adolescents, challenging the rules is age-appropriate and functional at times for emotional growth. It allows them to assert themselves as individuals and to begin to learn to take responsibility for their actions. However, for adolescents with FA this can be a time of rebelling against the "rules" of the disease. Young adults report stopping their medications, sun bathing, drinking alcohol, smoking, etc. Compliance with medication regimens may be of concern and should be given particular attention at this stage, as should the risk-taking behaviors associated with greater chances of malignancy.

As children get older, they need to be involved in assenting, consenting, and participating in actual decisions about their medical care. As their children become more active decision-makers, parents may feel some

relief that they are now making decisions with, rather than for, their children. Yet as children approach young adulthood, parents have expressed anxiety about how their children will learn to make complicated, sophisticated decisions for themselves. For some young adults, the decisions will continue to be made in partnership with their parents. Others will want full responsibility, and parents will need to trust their grown children's choices. This time of growth for the person with Fanconi anemia also becomes a time of growth for parents. There can be occasional dissonance between parents and children. In some cases, cognitive factors may limit the child's ability to make decisions.

Living with FA is a long and arduous journey for many children, yet they respond as children and often have more energy than adults would in similar circumstances. Children of all ages need to be allowed to continue to grow, regardless of the status of their medical conditions. Maximizing the capacity of the child with FA inherently helps all family members to acknowledge and delight in the child's gains, as opposed to focusing only on losses. Achievements, great or small, cultivate growth and satisfaction for both children and parents. Children need to be prepared to be successful and motivated in life, and not exclusively focused on Fanconi anemia. FA is a component of the life of the person who is diagnosed, but it is not what defines him or her.

## **Siblings**

Siblings present their own unique concerns, some visible and some invisible. They may feel guilty that the disorder happened to their sibling and not to them or may feel that they are less important because they are not getting as much attention. Siblings care about and

worry about each other a great deal. For many, their universe is defined by their role as either an older or younger brother or sister. Siblings of children with life-threatening or fatal illnesses often have as much of an emotional response to the illness as the affected children.

Open communication, the opportunity for expression, and the ability to process the experience help siblings to find their place in the world. It is important for families to address their unaffected children's feelings and questions, while involving them in the activities of the child(ren) with FA whenever possible. Siblings need their own time with parents, medical knowledge appropriate to their age, and to truly be and feel that they are an integral part of the family. Anxiety, jealousy, and guilt are among the emotions experienced by siblings.

The already complex relationships of siblings are further complicated when there is more than one child with FA in the family. Siblings often use each other as reference points, in life and beyond. These relationships have a very powerful presence that may not always be visible in a family. It is important that affected and non-affected siblings have the opportunity to talk with each other and with their parents. These can be among the strongest relationships in life and need to be cultivated and nurtured during this journey.

## **Young Adults and Adults with FA**

Becoming a young adult leads to a more comprehensive understanding of the illness, perhaps responding emotionally to Fanconi anemia in a new way, and addressing salient issues that may have been dormant at other developmental stages. Young adults who face the most severe manifestations of the illness may, of necessity,

remain more physically and emotionally dependent on family members. On the other hand, their family connections may reach deeper levels than those of their healthy peers. At each stage, issues of dependence and independence may need negotiation.

Advice to parents from a group meeting of young adults included:

- Don't worry about what is going to happen. It is going to happen anyway, so don't waste time.
- Don't feel guilty or responsible for the disease.
- Don't be overprotective.
- Don't forget the siblings.

Finding their own voices, taking responsibility for managing their own illness, becoming the primary decision-maker, using their parents as partners or consultants, and truly becoming independent are appropriate and very significant steps for young adults with FA. It is important to help such individuals gain their independence while helping them understand that they can still rely on their families for support and assistance. The partnership with parents should be well established long before this age. Family members need to work together to understand the best decision-making practices in their families. Fanconi anemia affects the whole family, not just when a child is initially diagnosed, but throughout the course of the illness. It affects the current generation and future generations. Some of the magnitude of the diagnosis is not apparent to the child until he or she reaches adolescence and young adult years.

Normal developmental challenges do not evade young adults with FA, yet age-appropriate experiences may have greater intensity and significance. Relationships,

peer pressure, experimentation with drugs and alcohol, and sexual relationships all pose emotional and physical challenges. Because of the inherent increased risks of cancers from some of these behaviors, young adults with FA (once informed) are torn between the desire to take care of themselves and their desire to fit in with peers. Ambivalence and anxiety can plague the young adult with FA, who needs to take care of himself or herself and constantly be on top of the unique challenges of living with the illness, while struggling to be like everyone else. It is difficult to understand how the multitude of illness-related factors affects the day-to-day emotional well-being and sense of self for persons with FA. Beyond the personal components of dealing with the disease, FA patients may feel accountable towards their peers with FA, parents, doctors, and the professionals with whom they have developed connections. This network may encourage young adults to do the “right thing.” Always present are the feelings of isolation and distance adults with FA may feel from those who do not have to face living with a life-threatening illness.

Deciding when to tell potential partners about FA—the short version, and then the long version—becomes part of the dating process for the person with FA. The issues of whom to tell, when to tell, and what to tell seem to be related to whom to trust and an ongoing evaluation of who needs to know what and why. These issues can frame early stages of relationships with roommates and romantic partners.

As relationships flourish, there is a natural inclination to think towards the future. This reflective process can be different for persons with FA who may simultaneously be trying to figure out their future goals in the context

of what they know about their medical condition. All of this may influence how they make choices of friends, relationships, careers, marriage, and parenthood.

Partners of young adults with FA often need help understanding the disease and its implications for their relationship, as well as the roles of other family members. Partners also need an outlet for information, expression, and help at times when their partner is not doing well or has to make major life decisions. Many understand the disease intellectually, but it is not until their partner's condition worsens that they begin to understand what some of their own concerns may be. Negotiating their roles as partners and with parents who have nurtured their children for decades can be quite challenging. Information, support, and counseling are important for this population.

For the adult with FA, having grown up with uncertainty of the future, establishing and mastering life goals, forging lifelong commitments, dealing with the issues of partnership, sexuality, marriage, children, ongoing cancer risks, financial and insurance concerns, and myriads of other problems all present unique challenges for this population.

These adult FA patients serve as an inspiration to all, yet should be recognized for their own needs, aspirations, and struggles. Increasing numbers of children are becoming young adults and adults with Fanconi anemia. In the same way that the needs of the children and then teens became a priority as treatment evolved, now the needs of these adults, physically and emotionally, become the priority. The medical course of Fanconi anemia is evolving, allowing for the emotional and physical sequelae to be better understood. Emotional connections for this group can be found in young adults

and adults with other rare illnesses who have survived to adulthood.

## **The Death of a Child**

If a patient nears death, the patient and the family need emotional support, clear thinking, concrete assistance, and tremendous understanding. By this point, the family has lived through many struggles with the illness and therefore may continue fighting longer than others might expect. Fighting, trying the next thing, and looking towards experimental options are the armor that families use to cope. For some, it may make sense to continue in that vein as long as possible. No one else should determine when a specific family should lose hope. Providing information and opportunities for discussion, helping families make decisions, supporting their choices, comforting, remembering, and remaining available are significantly helpful to families at this stage.

Support after the death of a child is necessary, but difficult to find. Rarely do bereaved parents feel that their loss is understood and therefore their ability to accept support, except from people in similar situations, may be limited. It is difficult to understand what they may be going through. Parental grief does not go away; it changes over time. Variables that have been shown to complicate mourning for families include: a markedly dependent relationship with the deceased; prior unresolved losses and stresses; a perceived sense of lack of support; death after an overly-lengthy illness; and the mourner's perception of preventability.<sup>2</sup> Families who lose a child to Fanconi anemia exhibit many of these factors. After having fought so hard, there can be a sense of guilt at not having been able to prevent the child's death.

Relationships with families should not end abruptly during the bereavement period, because it is a most difficult phase for them. Assisting families to understand many of the more intense feelings (anger, regret, loneliness, depression) as part of the natural process at this time is helpful. Ongoing communication to reflect on the child's life, referrals for counseling and support groups, and caring about the family's struggle are important. The death of one's child or one's sibling, regardless of the age of the child (young adults, older adults) is devastating, and can have lifelong implications for the family. The complication of having a genetic illness, an illness that a family will have to deal with for generations to come, adds to the complexity of coping after a child dies. FA will always be an issue for an affected family.

### **Recommendations for the Physician**

- Provide the opportunity for an initial psychosocial assessment of the child and family at the time of diagnosis.
- Provide the family access to appropriate counseling and other resources throughout the life of the person with Fanconi anemia.
- Provide developmentally appropriate communication for patients to enhance their understanding of and comfort with FA. Encourage dialogue among children with FA or other bone marrow failure diseases or other life-threatening illnesses.
- Encourage involvement with activities through the FA Research Fund to help families develop and maintain a current knowledge base, to gain support, and to afford families an active role in supporting research seeking to help their children.

- Encourage families to create a working partnership between the physician and the family, allowing for mutual respect for what each has to offer to the situation.

Enable patients, as they mature, to become responsible and proactive with regard to their medical care (recommended by a focus group of parents).

## References

1. Koocher GP, O'Malley JE. *The Damocles Syndrome: Psychosocial Consequences of Surviving Childhood Cancer*. New York, NY: McGraw-Hill; 1981.
2. Rando TA. The increasing prevalence of complicated mourning: the onslaught is just the beginning. *Omega* 1992-93; **26**: 43-45.

