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Fanconi Anemia

Fanconi anemia is the most common bone marrow failure syndrome. People with this disorder cannot produce normal white blood cells, red blood cells, or platelets. They also have an increased risk of developing myelodysplastic syndrome (MDS), leukemia, and/or solid tumors of the head and neck.

Children with Fanconi anemia may be shorter than others their age and have abnormal skin color, abnormal thumbs, and small eye size. Your child's care plan is tailored to his or her current health and needs and may include:

- blood transfusions
- growth factors: medicines that promote the growth of the blood cells your child is lacking
- immunotherapy, which revs up the body's own immune system
- chelation therapy: medicines that remove excess iron from the blood in patients who have too much iron

supportive care

a stem cell transplant: this uses stem cells donated by a healthy person to replace the abnormal bone marrow and help kickstart a healthy blood-forming system

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Congenital Amegakaryocytic Thrombocytopenia (CAMT)

Congenital amegakaryocytic thrombocytopenia (CAMT) is caused by a mutation in a gene called *MPL*. This mutation can lead to problems with red and white blood cells and platelets, and also raises the risk of leukemia. Children with CAMT can have bruising, bleeding, abnormal thumbs, and be shorter than their peers.

At MSK Kids, we treat patients with this disorder using allogeneic stem cell transplantation. This uses stem cells donated by a healthy, matched donor to replace your child's ineffective bone marrow. We have successfully performed transplants where we remove certain T cells from the donated marrow to reduce the risk of complications after transplant, and we give chemotherapy instead of radiation therapy before the transplant to avoid the high risks of radiation in young children. Your child's care includes lifelong monitoring and the expertise of other specialists to address any other health problems that may arise.

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Diamond-Blackfan Anemia (DBA)

Children with Diamond-Blackfan anemia (DBA) make small numbers of red blood cells. Their white blood cell and platelet levels are typically normal. This disorder is caused by mutations in genes that govern a cell's ability to produce certain proteins. People with DBA may have abnormal thumbs and be shorter than others their age.

When DBA only affects red blood cells — which is usually the case early in life — we can treat it with steroids such as cortisone and prednisone. These help stimulate the bone marrow to make red blood cells. Some children need blood transfusions every three to four weeks. If your child has a matched sibling who can donate stem cells, we can perform stem cell transplantation early in life. At MSK Kids, we have successfully treated young people with DBA. Your child's care includes lifelong monitoring and the expertise of other specialists to address any other health problems that may arise.

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Dyskeratosis Congenita

Dyskeratosis congenita causes problems with white blood cells, red blood cells, and platelets. More than half of the children with dyskeratosis congenita have a mutation in genes known to cause this disorder. In the remainder of cases, the genetic mutations are unknown. In addition to blood tests and exams, we use a highly sensitive screening test called telomere length analysis to help make the diagnosis. Children with dyskeratosis congenita may have darkened areas on the skin or abnormal skin color, nail changes, white patches in the mouth, abnormal thumbs, and small eye size, and be shorter than their peers.

We often treat children with dyskeratosis congenita using stem cell transplantation. Because we know that children with this disease have especially fragile skin, lungs, and livers, we take a less intensive approach that involves low-dose chemotherapy and the removal of certain T cells from the transplanted stem cells to reduce the risk of complications. This also makes the treatment more tolerable. When a child has dyskeratosis congenita, he or she is also at increased risk of other disorders, such as aplastic anemia, myelodysplastic syndrome, and acute myelogenous leukemia. We provide lifelong monitoring and connect you with other specialists to address any other health problems that may arise.

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GATA2 Deficiency

GATA2 deficiency is a rare genetic disorder caused by a mutation in the *GATA2* gene, which results in problems with the immune system. Children with GATA2 deficiency are prone to infections, respiratory problems, hearing loss, and leg swelling. They also have a higher risk of certain blood cancers, such as myelodysplastic syndrome and leukemia.

Treatment for GATA2 deficiency often includes medications and other therapies to manage the complications of the disorder, such as antibiotics for bacterial infections and treatment of leg swelling. A stem cell transplant from a matched donor is the most effective therapy. Your child's team will let you know the best treatment approach. We also provide lifelong monitoring and connect you with other specialists your child may need.

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Shwachman Diamond Syndrome

Shwachman Diamond syndrome begins by affecting the production of white blood cells, then progresses to creating problems with red blood cells and platelets. Almost all children with Shwachman Diamond syndrome have mutations in a gene called *SBDS*. They may also have bone abnormalities and problems with the liver and pancreas, which can interfere with fat absorption in the body. Children with Shwachman Diamond syndrome may also have abnormal skin color, abnormal thumbs, and small eye size, and be shorter than others their age.

To treat Shwachman Diamond syndrome, doctors at MSK Kids usually start with supportive care to help pancreatic issues and blood transfusions to boost red blood cell and platelet counts. If your child develops severe aplastic anemia or myelodysplastic syndrome, we'll likely recommend a stem cell transplant. We take a less intensive approach that involves low-dose chemotherapy and the removal of certain T cells from the transplanted stem cells to reduce the risk of complications. This also makes the treatment more tolerable. Because Shwachman Diamond syndrome can affect multiple organs and raise the risk of other health problems, we provide lifelong monitoring and connect you with other specialists your child may need.

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Thrombocytopenia Absent Radii (TAR Syndrome)

Children with TAR Syndrome are missing one of the two bones (known as the radius) from each lower arm. This disease is caused by a mutation in a gene called *RBM8A* and leads to low levels of platelets. Children with TAR Syndrome are also more prone to bruising. If your child has TAR Syndrome, we typically start with supportive care, such as medications. In rare occasions, we may be able to perform a stem cell transplant. Because TAR Syndrome can raise the risk of other health problems, we provide lifelong monitoring and connect you with other specialists your child may need.

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