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Welcome –

We are happy that you found us in your journey with Shwachman-Diamond Syndrome. We have gathered together this information for new families and believe that you will find it useful.

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All of the information provided in this packet is for informational purposes only. We do not recommend any particular product or treatment protocol. Please discuss this information with your physician to confirm that it applies to you or your child.

For further resources, please visit our website at www.shwachman-diamond.org.

If there is anything we can do to help you, please feel free to call toll free at 1-888-825-SDSF (7373) or email us at info@shwachman-diamond.org.

What is Shwachman Diamond Syndrome?

Shwachman Diamond Syndrome was first identified in 1964. It involves many systems in the body, including the pancreas, bone marrow and skeleton.

CAUSE

SDS is caused by an autosomal genetic mutation. This means that a gene mutation is provided from each parent. The genetic mutation that causes SDS is located on the long arm of chromosome 7. However, about 10% of patients have symptoms of SDS but do not have a known genetic mutation.

EARLY SIGNS

Infants with SDS usually show symptoms early. They may have feeding problems, failure to thrive (do not gain weight), do not grow and may have frequent infections. They may also have skeletal abnormalities.

WHAT BODY SYSTEMS ARE INVOLVED?

Many body parts are affected by SDS among them are the bone marrow, pancreas, skeleton, kidneys, liver, brain, teeth and immune system.

PANCREATIC ISSUES

The pancreas is an organ that has two functions. It produces insulin to control blood sugars in the body. It also produces enzymes to digest food and nutrients. In patients with SDS the part of the pancreas that produces enzymes does not work properly. This results in greasy, frequent and foul smelling stools. Enzyme replacement will help the patient digest food and provide needed nutrition. Because fats are not easily digested attention should be given to monitoring the levels of the fat soluble vitamins (A,D,E and K) and replacement supplements provided when necessary.

In about 50% of patients pancreatic function improves with age and enzyme replacement therapy may be discontinued. Enzymes should only be discontinued after consultation and confirmative testing by a gastroenterologist. Slow improvement in weight is usually seen but growth velocity may not improve.

Some patients also have issues with blood sugar levels.

BONE MARROW

The bone marrow produces all lines of blood cells: white cells, red cells and platelets. In SDS patients the bone marrow does not function properly. As a result, the patient may have decreased numbers of white cells, red cells and/or platelets.

ANEMIA

Anemia refers to a decreased number of red blood cells in the blood stream. Red blood cells are needed to carry oxygen to the cells in the body.

NEUTROPENIA

Neutropenia is a condition caused by a reduction in the number of white blood cells called neutrophils. These cells are responsible for helping the body fight bacteria and fungal infections. Patients with an absolute neutrophil count (ANC) that is low (below 1500 per microliter) may be at risk for life threatening infections.

THROMBOCYTOPENIA

Blood platelets clot the blood and prevent bruising. A normal platelet count is above 150,000 per microliter. A decreased number of these cells is called thrombocytopenia. If the platelet count is too low, platelet transfusions may be necessary to prevent hemorrhaging.

It is strongly recommended that every SDS patient be seen regularly by a hematologist, a physician who specializes in the blood issues. Patient's blood counts should be regularly monitored. It is further recommended that all patients have periodic bone marrow biopsies to monitor how the bone marrow is functioning.

Further complications that can develop are leukemia, myelodysplastic syndrome (MDS) and aplastic anemia. These complications are life threatening and require immediate care by specialists who are experienced in treating SDS patients.

SKELETAL

Bone abnormalities are reported in some patients. These usual involve the hips, rib cage, femur and tibia. If severe enough they may require surgical intervention.

LIVER

An enlarged liver and elevated liver enzymes is frequently seen in young patients. Liver enzyme levels do normalize with age. Chronic liver disease has been documented.

NEURO/PSYCH ISSUES

Developmental delays and learning difficulties are seen in some SDS patients. These may include learning disabilities, developmental delays, ADHD and behavior issues. Testing is recommended if there is indication of these issues.

OTHER POSSIBLE COMPLICATIONS

Less common features include dental dysplasia and increased dental caries, lung disease, testicular fibrosis and cardiac lesions.

SDS is a very rare condition. For this reason, not many physicians are knowledgeable about it. We feel that the best possible care for your child can be obtained by taking him/her to physicians who have knowledge and experience in caring for SDS patients. We are happy to help you find a specialist.

Understanding the Blood

In order to understand the blood problems, which are sometimes associated with Shwachman-Diamond syndrome, it may be helpful to learn about normal blood cells and what healthy blood is like.

We all know that blood is essential to life. Blood carries oxygen, nutrients, hormones and chemicals to cells throughout the body. Blood plays an essential part in protecting the body from infection. Blood also helps remove waste and toxins from the body.

Blood is made up of many different cells. The three main kinds of blood cells are:

Red blood cells contain hemoglobin. Hemoglobin is an iron-rich protein. Oxygen is picked up by the hemoglobin as it passes through the lungs. The oxygen is carried by the red blood cells and given to different organs and body tissues. When the red blood cell count is low in number, it is anemia. A person who is anemic may feel dizzy, short of breath and have headaches because they do not have enough oxygen in their blood.

Platelets are small disc-shaped cells that help blood clot. Platelets prevent abnormal or excessive bleeding. If there are not enough platelets, serious bleeding and/or bruising can result.

White blood cells defend the body against infection. There are three types of white blood cells. Each has its own infection-fighting job.

1. **Monocytes** defend the body against some bacteria, such as tuberculosis.
2. **Granulocytes** are divided into three types: neutrophils, eosinophils and basophils. **Neutrophils** fight infection by quickly increasing in number, then engulfing and killing microorganisms. The neutrophils then quickly return to their pre-infection number. They fight infection caused by bacteria and fungi. **Eosinophils** and **Basophils** also have a role in infection fighting.
3. **Lymphocytes** are divided into two cell types which work with the immune system. **T-cells** attack viruses and cancer cells and control the function of other white cells. **B-cells** make and release antibodies. Antibodies attach to infectious agents and mark them for removal from the body.

A decrease in any type of white blood cell (WBC) may result in an increased risk of developing infection. In patients with Shwachman-Diamond syndrome it is most often the neutrophils that are low in number.

There are two additional factors which play a role in Shwachman-Diamond syndrome.

Neutrophils make up between 42 and 72% of the total number of white blood cells. In healthy persons, this count stays relatively consistent. In the absence of an infection, it always stays at about the same number.

In some Shwachman-Diamond patients, the percentage of neutrophils can fluctuate between the normal range and low. This condition is called ***cyclic neutropenia***. When the percentage of neutrophils dips below normal, the patient is more susceptible to infection. The low cycles occur about every 21 days and may last for 3 to 6 days.

In a smaller number of Shwachman-Diamond patients, the percentage of neutrophils is always lower than normal. This can result in an even greater susceptibility to infection.

Most experts feel that as many as 95% of patients with Shwachman-Diamond syndrome are affected by either cyclic or constant neutropenia.

Also, as stated above, the way that neutrophils defend the body from infection is by quickly increasing in number and rushing to the site of an infection, surrounding and destroying the bacteria and/or foreign substance. In some Shwachman-Diamond patients, a condition also exists called ***neutrophil immobility***. When neutrophil immobility is present, the neutrophils do not function normally. They do not move as quickly and efficiently to the site of an infection. This is an additional deterrent to the body's ability to fight bacteria infection.

This information is provided *only* as a guideline. We urge you to discuss this material with your child's physician to see if it is relevant to your child.

This material was produced by Shwachman-Diamond Syndrome Foundation. We gratefully acknowledge Susan Burroughs, MD for reviewing this material for us.

Determining your ANC (Absolute Neutrophil Count)

A Complete Blood Count (CBC) also known as a Full Blood Count (FBC) measures the levels of the three basic blood cells-white cells, red cells, and platelets.

An ANC (Absolute Neutrophil Count) measures the percentage of neutrophils (shown in this listing as Polys) in your white blood count.

multiply your white blood count (WBC) x total neutrophils (segmented neutrophils% + segmented bands%) x 10 = ANC.

A normal ANC is over 1,000. An ANC of 500-1,000 is considered neutropenic and the Registry considers that an individual whose ANC is chronically less than 500 has Severe Chronic Neutropenia.

Result column: shows counts that fall within the normal range.

Flag column (this marks items that are out of range): shows counts that are lower ("L") or higher ("H") than the normal range.

White blood cells: help protect you from infection.

For this patient, the total white cell count is 2.0, which is low.

Reference Interval (or Reference Range) column: shows the normal range for each measurement. Different labs may use different ranges, your test results may be slightly different, depending on where your results are processed.

Red Blood Cells: Carry oxygen from your lungs to the rest of your body.

Hemoglobin (Hb or Hgb): the part of the red cell that carries the oxygen.

Hematocrit (HCT), is a measure of the amount of red blood cells in the blood.

Platelets: the cells that form blood clots that stop bleeding. The platelet count for this patient is normal.

Polys (also known as segs, segmented neutrophils, neutrophils, granulocytes) are the most numerous of our white blood cells. These are the first line of defense against infection, killing invaders of the body.

Bands (also known as stabs, segs or segmented bands) are immature polys. They also function to kill invaders of the body.

Lymphs or lymphocytes are white blood cells which assist in building immunity and include B and T cells.

Monocytes, eosinophils, and basophils destroy invading bacteria and viruses.

Differential: part of the CBC that shows counts for the five main kinds of white cells, either as percentages (the first 6 counts), or as the number of cells (the second 6 counts). This patient has a lower than normal **poly** count and a higher than normal **lymph** and **monocyte** count.

Test	Result	Flag	Units	Reference Interval
CBC WITH DIFFERENTIAL				
White Blood Count	2.0L		x 10 ³ /μL	4.8-10.8
Red Blood Count	4.34L		x 10 ⁶ /μL	4.70-6.10
Hemoglobin	13.2L		g/dL	14.0-18.0
Hematocrit	37.5L		%	42.0-52.0
Platelets	278		x 10 ³ /μL	130-400
Polys	14.8L		%	43.0-65.0
Bands	5		%	
Lymphocytes	55.5H		%	20.5-45.5
Monocytes	22H		%	5.5-11.7
Eosinophils	1.7		%	0.9-2.9
Basophils	1.0		%	0.2-1.0
Atypical lymphs	0.0		%	0.0-2.0
Polys (absolute)	0.3L		x 10 ³ /μL	2.2-4.8
Bands (absolute)	0.1		x 10 ³ /μL	
Lymphs (absolute)	1.1L		x 10 ³ /μL	1.3-2.9
Monocytes (absolute)	0.4		x 10 ³ /μL	0.3-0.8
Eosinophils (absolute)	0.0		x 10 ³ /μL	0.0-0.2
Basophils (absolute)	0.0		x 10 ³ /μL	0.0-0.1
Atypical lymphs (absolute)	0.0		x 10 ³ /μL	0.0-2.0

To determine your ANC:

1. Find the WBC, the polys and bands on your CBC.
WBC 2.0
Polys 14.8%
Bands 5%
 2. Add the polys and bands.
(14.8 + 5 = 19.8)
 3. Multiply the sum of the polys and bands by the WBC.
19.8 x 2.0 = 39.6
 4. Multiply the product by 10.
39.6 x 10 = 396
- This person has an ANC of 396**

To calculate the ANC from absolute numbers the formula is:
Absolute polys + Absolute bands multiplied by 1000 = ANC
(0.3 + 0.1) x 1000 = 400

THE USE OF PANCREATIC ENZYMES IN SHWACHMAN-DIAMOND SYNDROME

A Guide for Parents and Patients

Proper nutrition is an important part of the treatment of Shwachman-Diamond syndrome. It is essential for normal growth and development.

In the healthy body, digestive enzymes are produced by the pancreas and secreted into the intestine. Enzymes digest or break down the foods we eat into their smallest components. The body then absorbs these components and uses them for energy and growth.

In most patients with Shwachman-Diamond syndrome, the pancreas cannot produce enough enzymes to digest food properly and so many components of the food cannot be absorbed. This results in poor nutrition and growth. To overcome this problem, pancreatic enzymes are taken orally.

TYPES OF ENZYMES

Enzymes are available as capsules in two forms: as powder and as enteric-coated microspheres (ESC). The powder is used by some doctors for infants. The capsules may be broken open and the contents mixed with a suitable food, such as applesauce. ECS capsules are effective for most other patients. The coating of the microspheres protects the enzymes from being destroyed by the acid in the stomach. ECS capsules can also be opened and the enzymes mixed with a food (usually applesauce) for those unable to swallow the capsules.

ENZYME DOSE

The amount of enzyme required varied from one person to another. The following table is a general guide to dose.

Amount of enzyme required when 8,000 unit lipase powder capsules and 8,000 units of lipase enteric coated capsules are used.

AGE	TYPE OF FOOD	ENZYME DOSE	
		Powder capsules (8,000 units per capsule)	ESC capsules (8,000 units per capsule)
0-12 mos.	Formula or homogenized milk	1 per 4 oz.	1 per 8 oz.
	Solids; cereal, meats or vegetables	1 per ½ jar strained food (1/2 jar = 4 tbsp)	1 per jar strained food (1 jar = 8 tbsp)
1-4 years	Meal	4-5	2-3
	Snack*	2-3	1-2
5-12 years	Meal	6-10	3-5
	Snack*	2-6	1-3
Over 12 yrs.	Meal	10-16	5-8
	Snack*	4-6	2-3

*Dose depends on the size and type of snack

THE USE OF PANCREATIC ENZYMES (CONTINUED)

QUESTIONS YOU MAY ASK

Q When during the meal should enzymes be taken?

A It is best to take enzymes throughout the meal. They may be divided into three equal portions. One-third should be swallowed at the start of the meal, another third halfway through and the rest near the end of the meal. Some people find that taking enzymes at the beginning and in the middle of the meal works well; for example, if six enzymes are required, take four at the beginning and two in the middle of the meal.

Q How will I know if the enzyme dose is correct?

A Signs of incorrect dosage are loose, frequent stools and abdominal pain. If frequent, foul-smelling stools persist, the doctor, dietician or nurse should be informed.

The most accurate way to evaluate the enzyme dose is by a 3-day stool collection in which the amount of unabsorbed fat is measured. Your doctor or dietician will let you know if this test is required.

Q What foods do not require enzymes for digestion?

A Foods that contain only sugar may be eaten without enzymes. These include all fruits, fruit juices and fruit drinks (lemonade, Kool-Aid, Quench, etc.) soft drinks, Jell-O, plain candies (not those containing chocolate or nuts) and popsicles.

QUESTIONS PARENTS OF INFANTS ASK

Q Will I be able to breast-feed my baby?

A Breast-feeding is encouraged for infants with Shwachman-Diamond syndrome. If the baby's weight gain is not adequate, fortified breast milk or concentrated formula may have to be offered by supplement your baby's intake.

Q What should I watch for when my baby starts taking enzymes?

A Diaper rash, mouth ulcers and skin irritation the mouth may develop during the first month your baby takes enzymes. These problems become less bothersome with time.

To minimize skin irritation, apply Vaseline generously around your baby's mouth before feeding. In addition, wiping your baby's mouth out with a wet sponge or cloth after enzyme administration will help.

Q What should I mix the enzymes with?

A You can mix the enzymes with almost any food. We have found that mixing enzymes with applesauce works well and camouflages the unpleasant taste of the powder enzymes. However, once you mix enzymes with the food, it must be eaten immediately. Never allow food in which enzymes have been mixed to stand for more than 2 or 3 minutes. Any delay in eating leads to breakdown of the food by the enzyme and in many cases destruction of the enzyme by the acid in the food.

Always mix enzymes with the same food. This will mean that your child will associate only that food with medication.

THE USE OF PANCREATIC ENZYMES (CONTINUED)

Do not mix enzymes in the bottle because they coat the bottle and begin to digest in the formula. However, if your baby is refusing the applesauce, you may mix the enzymes in a small amount of liquid, either formula or juice, and feed your baby with a spoon or dropper.

If your child is old enough to drink from a straw, you may try to use the straw-method to administer enzymes. Simply put water in a cup and use a “bendable” straw. Pinch the top of the straw with your fingers and dump the enzymes in the top of the straw (the enzyme beads will not fall down the straw because your finger is clamping the straw at the top). Have your child start sucking the water up from the straw. When the child sucks the water, let go of your pinch around the straw. You may have to practice this approach until it feels comfortable for both you and the child.

Q How much enzyme should I give when my baby first starts eating solid foods?

A Usually babies take only a few teaspoons of food when they first start eating solids. At this point, they do not usually need any more enzyme than they are already taking with their breast milk or formula.

When your baby is eating $\frac{1}{4}$ cup or $\frac{1}{2}$ jar of strained food, you should increase the amount of enzyme he/she is receiving. See guidelines in the table “Enzyme Dose”.

*Adapted from “The Use of Pancreatic Enzymes in Cystic Fibrosis” Hospital for Sick Children by:
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Fat-Soluble Vitamin Supplementation

Introduction

The fat-soluble vitamins (A, D, E, and K) as the name implies, are found mainly in various forms of dietary fat. Supplementation with these vitamins is usually unnecessary, unless an individual has fat malabsorption, or trouble digesting the fat in the diet. This is the case with people who have Shwachman-Diamond syndrome, cystic fibrosis or other diseases of the pancreas. Oral pancreatic enzymes may be required by some individuals with Shwachman-Diamond syndrome, improving fat absorption considerably, but not enough to ensure adequate intake of the fat-soluble vitamins (the most susceptible vitamins to fat malabsorption are vitamins E and K). This is the reason fat-soluble vitamins are prescribed as an oral vitamin supplement. It is advisable to take a water miscible preparation which may be better absorbed than an oily one. However, an intake of very large doses of fat-soluble vitamins, especially vitamins A and D, can result in serious health consequences.

VITAMIN A

Vitamin A has many important functions in the body, including its role for night vision and adaptation to light. If vitamin A deficiency occurs over a period of years, then night blindness or a decreased ability to adapt to darkness can occur. Sufficient vitamin A can cure night blindness. Vitamin A plays a role in maintaining healthy skin and in the body's ability to fight infection. It is also necessary for normal growth and development. The daily requirement for vitamin A is 400 RE (1300 IU) for infants and 400-1000 (1300-3300 IU) for children and adults. Food sources of vitamin A include eggs, liver, tomatoes, milk and some fruits and vegetables. The liver is able to store vitamin A, so supplementation is usually unnecessary, unless fat malabsorption is present. For individuals with fat malabsorption, supplementation with the recommended daily intake (~ 1500 IU/day for infants, 1500 – 4000 IU/day for children and adults) is advised.

If excess vitamin A is offered, at just 10 times the recommended daily intake over a period of weeks to years, toxic symptoms can appear. These range from bone loss, loss of appetite, headache, dry skin and hair loss to vomiting and liver enlargement.

VITAMIN D

Vitamin D is a vitamin and a hormone (when the skin is exposed to sunlight, vitamin D is formed; there it is also described as a hormone). Vitamin D is absorbed with dietary fat in the intestines. It is transported to the liver and then the kidney. Once activated, it helps reabsorb calcium and release it from bone when calcium levels are low. It helps improve absorption of calcium and phosphorus, which are important for bone development. Without adequate amounts of vitamin D, bones can weaken. In children, rickets can result and in adults, osteomalacia, which means soft bones. Fractures of the hip and spine and other bones can occur. The recommended daily intake of vitamin D is 400 IU/day for children and adults. While foods such as eggs and cereals contain some vitamin D, the main dietary source is milk. For individuals with fat malabsorption, supplementation with 400 IU/day is recommended.

Fat-Soluble Vitamin Supplementation Continued

Vitamin D (continued)

If even only 5 times the recommended daily intake is ingested regularly, then vitamin D toxicity can result. Over absorption of calcium can result with calcium deposits on the kidneys and other organs. Weakness, loss of appetite, diarrhea, vomiting, mental confusion and increased urine output can also occur.

Vitamin E

Vitamin E has gained popular attention for its role as an antioxidant. Many enzyme systems produce potentially damaging oxidative products and the body has a complex system of defense (including vitamin E) against them. This means that vitamin E can help cells protect their outer membranes and prevent their destruction. A deficiency of vitamin E can cause breakdown of red blood cells, especially in infants, resulting in anemia. Vitamin E is also necessary for the maintenance of the immune system and nervous tissue. Deficiency of vitamin E can result in neurological disorders affecting the spinal cord and retinas. The recommended daily intake of vitamin E is 4-5 IU/day for infants and 6-15 IU/day for children and adults. Plant oils are rich in vitamin E, as are margarine and some fruits and vegetables. Grains such as oatmeal and wheat germ are also good sources. In individuals with fat malabsorption, 50 IU/day for infants and 100-200 IU/day for children and adults are usually prescribed to supplement dietary intake.

Vitamin E is not very toxic. However, if 50 times the recommended daily intake is consumed, nausea, weakness, headache, diarrhea and fatigue can occur.

Vitamin K

Vitamin K plays an important role in blood clotting. About one half of the daily intake of vitamin K comes from the diet and the other half is produced by the bacteria in the intestines. Vitamin K is found in many green vegetables and in liver. The recommended daily intake is 5 micrograms/day for infants and 15-80 micrograms/day for children and adults. The amount in our diet alone provides antibiotics (bacteria in the intestines may be destroyed) and in those with fat malabsorption, vitamin K deficiency can occur. Signs of deficiency including poor malabsorption require daily supplementation of at least 2 times the recommended daily intake.

Vitamin K toxicity is rare. If excess amounts of vitamin K are given, an individual may become jaundiced or develop a certain type of anemia.

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CARE OF MOUTH SORES

Many children and adults with Shwachman-Diamond syndrome get sores in their mouth as the result of neutropenia. Neutropenia is said to affect about 95% of people with Shwachman-Diamond syndrome, at one time or another. These mouth sores frequently bother some Shwachman-Diamond patients, while others get them infrequently.

The following is a list of suggestions to ease the pain and promote healing.

- *Use **only** a soft bristled toothbrush. Your doctor may even recommend using a disposable foam stick or cotton swab instead of a toothbrush.
- *Rinse mouth every few hours with a normal saline solution (1 teaspoon of salt in a quart of water).
- *Use a nonirritating cleanser such as baking soda.
- *Avoid dental floss if it causes pain and/or bleeding.
- *Avoid commercial mouthwashes, as they can irritate the mucous membrane. You may want to talk to your doctor about using a prescription mouthwash.
- *Avoid alcohol and tobacco, which can also irritate the mucous membrane.
- *If standard toothpaste cause irritation, try bubble gum flavored children's toothpaste.

IF IT HURTS TO EAT

- *Continue rinsing with a saline solution.
- *Cold food, such as popsicles, may help ease the pain.
- *Eat soft or liquid foods.
- *Experiment with food temperatures to see what temperature is most comfortable. usually chilled foods and foods served at room temperature are preferred. Hot foods should be avoided.
- *Avoid highly seasoned and spicy foods, citrus fruits and juices and carbonated beverages.
- *Avoid coarse foods, such as dry toast, chips and crackers. Chips and crackers may also be dusted with salt, which can cause pain.

Please remember that this is only a guideline. All medical problems should be discussed with your physician.

Sick Kids researchers identify gene for Shwachman-Diamond syndrome

TORONTO (December 23, 2002) — Researchers at The Hospital for Sick Children (HSC) and the University of Toronto (U of T) have identified the gene that is altered in Shwachman-Diamond syndrome. The researchers studied 250 Shwachman-Diamond syndrome families from around the world and identified two major disease-causing mutations in a gene on chromosome 7. This research is reported in the January issue of the scientific journal *Nature Genetics*.

Shwachman-Diamond syndrome (SDS) is a relatively rare genetic disorder that occurs in approximately one in 50,000 births. SDS affects many organs in the body. Primary features of SDS include a defect in the pancreas that leads to difficulties in digesting food, hematologic (blood) problems with inadequate production of some types of white blood cells, skeletal abnormalities, and short stature. The hematologic problems make people with SDS prone to severe, sometimes fatal infections, and some die from blood complications such as leukemia or bone marrow failure.

“The identification of the gene is important because it will allow for accurate diagnosis and screening of Shwachman-Diamond syndrome. It will also help us to determine what goes wrong at the molecular level, and this will open the door to the development of new therapies,” said Dr. Johanna Rommens, the study’s principal investigator, an HSC senior scientist and associate professor of Molecular and Medical Genetics at U of T.

“This discovery will aid in the clinical management of Shwachman-Diamond syndrome,” said Dr. Peter Durie, co-principal investigator of the study, an HSC gastroenterologist and senior scientist, and a professor of Paediatrics at U of T. “It is also very important to the families affected by this disease. We have received patient samples from around the world, and the Shwachman-Diamond parent groups from many countries supported this research financially.”

Shwachman-Diamond syndrome is an autosomal recessive disease, meaning that a child needs to inherit two mutated genes (one from each parent) in order to have the disease. The SDS gene resides in a region of the human genome that was very difficult to map because it contains a lot of highly repetitious DNA sequence. It was found that in the normal state, every chromosome 7 has two copies of the SDS gene — a functional gene and a non-functional gene relic, called a ‘pseudogene’.

“We have determined that the type of genetic mutation that causes Shwachman-Diamond syndrome is gene conversion, in that a piece of the non-functional pseudogene has been introduced into the good copy of the gene, thus disrupting its function. These types of mutations have been seen in more than 90 per cent of SDS patients,” said Graeme Boocock, the

study's lead author and a University of Toronto graduate student. Boocock is a recipient of a Canadian Institutes of Health Research doctoral research award.

Other members of the research team are Jodi Morrison, Maja Popovic, Nicole Richards, and Lynda Ellis, all from The Hospital for Sick Children.

This research was supported by the Canadian Institutes of Health Research, Shwachman-Diamond Syndrome Canada, Shwachman-Diamond Syndrome International, Shwachman-Diamond Support of Great Britain, The Harrison Wright Appeal, Shwachman-Diamond Syndrome Support of Australia, Paediatric Consultants Inc., Canadian Genetic Diseases Network of Centres of Excellence, and The Hospital for Sick Children Foundation.

The Hospital for Sick Children, affiliated with the University of Toronto, is Canada's most research-intensive hospital and the largest centre dedicated to improving children's health in the country. Its mission is to provide the best in family-centred, compassionate care, to lead in scientific and clinical advancement, and to prepare the next generation of leaders in child health. For more information, please visit www.sickkids.ca.

Further information for families affected by Shwachman-Diamond syndrome is available on the HSC Web site at www.sickkids.ca/mediaroom/custom/SDSgeneQA.asp.

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Shwachman-Diamond Syndrome

Gene Questions and Answers

With participation of SDS families from around the world, researchers from the Dr. Johanna Rommens and Dr. Peter Durie laboratories at The Hospital for Sick Children in Toronto have identified the altered gene that causes this disorder in December of 2002.

Does a genetic test for Shwachman-Diamond Syndrome (SDS) exist?

The gene that causes Shwachman-Diamond syndrome has been identified and a number of genetic mutations (mistakes in the gene) have been found in people who have this condition. A genetic test is now available and is available at many testing centers. Please refer to the "Diagnosis Information" section of the website for a list of the testing centers.

(Additional information from the SDS Registry, which is not part of this original Q&A, states that "Genetic testing of siblings in families of patients with Shwachman-Diamond syndrome is important and necessary. A recent study through the North-American Shwachman-Diamond Syndrome Registry looking at initial symptoms at the time of diagnosis reports that as many as half of patients with SDS may first see a doctor without the classic symptoms of neutropenia, diarrhea or poor growth/failure to thrive. These results included some siblings of patients with Shwachman-Diamond syndrome who were otherwise without symptoms, and who were diagnosed with the SDS upon genetic testing. **All siblings of patients with SDS should be tested**, as timely diagnosis prior to the development of complications is critical for the best medical management and outcomes, particularly in the case of bone marrow failure, leukemia, or bone marrow transplantation. This is also especially important to ensure that affected siblings are not used as donors for bone marrow transplantation.")

What is the difference between genetic testing in a research lab and a "routine clinical test"?

Research-based genetic testing is different from routine clinical tests. Research procedures are not approved nor standardized, and do not involve the same level of quality control that is required by regulatory authorities for commercial laboratories. The results of research-based tests include a disclaimer stating that the findings do not involve a standardized, approved test, and a caution that there may be errors in the test results.

When will a test be available for my spouse to see if he/she is a carrier?

A genetic test for carrier status is available at the Molecular Genetics Laboratory at The Hospital for Sick Children, Toronto, Canada. Please contact info@shwachman-diamond.org for the current contact information.

Can you test siblings to determine their carrier status?

Current knowledge suggests that people who carry a single SDS mutation do not have clinical symptoms.

Is there more than one gene mutation?

To date, all SDS-associated mutations have been identified in one gene. There are a number of different mutations within this one gene. Two common alterations were found to account for 75% of all mutations in the initial group of families we have tested.

What does identification of the SDS gene mean for the short-term?

In the short term, discovery of the SDS gene will lead to improved diagnosis of the disease since genetic tests are become available. In addition, collected clinical and genetic data can be used to determine if some mutations are associated with distinct SDS features.

What is next for research?

The focus of research will now shift towards studying the gene that is altered in SDS. We need to learn more about this gene, its role in cells and tissues, how it works, and what goes wrong in SDS. This insight will be crucial to learn how mistakes in this gene lead to the clinical symptoms of SDS, including blood, bone and digestive problems. Ultimately, this may lead to development of new therapies for SDS. It is now widely accepted that SDS falls in the category of a ribosomal disease meaning that the ribosome is affected.

Will finding the gene mean that there is a "cure" (gene therapy)?

Intense research efforts worldwide are focused at correcting genetic defects by gene therapy. However, there are numerous obstacles and safety concerns about introducing a corrected gene into human cells. We hope that breakthroughs in this field will make this an option for SDS patients in the future.

Why must I go through my physician to receive this genetic information?

Issues arising from genetic test results can be very complex. Your physician can help explain the results and prepare you for the possible implications of having this information. Your doctor may also decide to refer you to a genetic counselor for additional consultation.

Is genetic counseling important?

The purpose of genetic counseling is to answer questions you may have and also to prepare you to deal with the results of genetic testing. It is important that you fully consider the risks and benefits related to genetic testing. It may not provide you with clear and final answers regarding the health of your child or other family members and the results can be inconclusive. There are other potential risks of learning about gene alternations in your family that cannot be predicted.

There may be unknown personal, legal and social consequences, and there may be implications for obtaining life and medical insurance.

When will my doctor receive the results?

Genetics research is complex and time-consuming. There may not have any findings for a small percentage of patients. In some cases, this will be due to inadequate amounts of DNA. In these cases, you will be requested to send an additional blood sample in order to complete genetic testing. Families who fit into this category will be contacted to discuss further testing.

Will I need to have my research results confirmed in a registered diagnostic facility?

Yes. This is particularly important if the genetic test results are to be used for genetic counseling, or if the clinical diagnosis is in doubt. This should be discussed with your doctor or a genetic counselor.

If mutations are not identified, does this mean I don't have SDS?

A diagnosis of SDS cannot be ruled out based on lack of genetic evidence. This is because some types of genetic mutations are rare and/or very difficult to detect. Genetic testing does not replace the need for careful investigation and monitoring of clinical symptoms, including pancreatic function and hematological problems.

Thanks to these dedicated doctors and Fellows below who wrote and answered these important questions above.

Graeme R.B. Boocock
Lynda Ellis, R.N.
Nicole Richards
Jodi Morrison

Research Study Opportunities

NORTH AMERICAN SDS REGISTRY

SDSF encourages every patient with SDS to register in the Shwachman Diamond Syndrome Registry. The more data that is available to doctors and researchers, the faster they can help with diagnosis, treatments and hopefully a cure!



The North American Shwachman-Diamond Syndrome Registry (SDSR) was established to collect medical information and clinical samples on all individuals with SDS with the goal of improving diagnosis and treatment.

The **SDS Registry** is dedicated to gathering and analyzing information about SDS and sharing any new knowledge with the SDS community and medical professionals. There is no charge to participate in this study. Participating in the study will help the SDSR better understand SDS and may eventually lead to better diagnostic tests and new forms of treatment.

To participate in the SDS Registry, please contact one of the following:

***Dana-Farber Cancer Institute/
Boston Children's Hospital***

Research Nurse

Maggie Malsch, RN

Phone: 617-355-4685

Maggie.Malsch@childrens.harvard.edu

***Cincinnati Children's
Hospital Medical Center***

Research Nurse

Sara Loveless, RN

Phone: 513-803-7656

sara.loveless@cchmc.org

For more information, please visit www.sdsregistry.org.

Research Study Opportunities

NCI IBMFS COHORT STUDY

Inherited bone marrow failure syndromes (IBMFS) are rare disorders in which there is usually some form of aplastic anemia (failure of the bone marrow to produce blood), associated with a family history of the same disorder. Some of these conditions have typical changes in physical appearance or in laboratory findings which suggest a specific diagnosis. There are several well-described syndromes, which can be recognized by health care experts. There are also patients who are harder to classify, but who appear to belong in this category.

Patients with these syndromes have a very high risk of development of cancer (either leukemia or certain solid tumors). At the moment we cannot predict which specific patient with an IBMFS is going to develop cancer. The **NCI IBMFS Cohort Study** enrolls North American families in which at least one member has or had an IBMFS. The study:

- includes individuals known to have an IBMFS as well as their first degree relatives (brothers, sisters, parents, and children);
- collects clinical information from study participants and their physicians;
- performs detailed physical examinations, x-rays and routine laboratory tests on those who are interested in traveling to the NIH to be seen in person by our team;
- attempts (on a research basis) identification of the specific genetic mutation that is associated with each family's disease;
- screens participants for early changes related to the specific cancers that occur in each syndrome;
- performs detailed research laboratory studies on blood and tumors collected from study participants, in an effort to understand the process by which cancers develop;
- monitors study participants in an ongoing fashion to determine the rate at which complications develop related to each disease, and to identify those complications more precisely;
- provides suggestions to study participants and their physicians regarding how to best take care of family members who are affected with a particular IBMFS; and
- offers genetic counseling, and an opportunity to learn the results of mutation testing, for those persons who decide that this information will be of use to them.

To enroll or inquire about this study, please call 1-800-518-8474.

The Principal Investigator responsible for this study is **Blanche P. Alter, MD, MPH**.

Their overall goal is to reach a better understanding of how cancers develop in persons with IBMFS, so that they may improve the health care which can be offered to persons with these disorders.

For more information, please visit: <http://www.marrowsfailure.cancer.gov/>

SDS Research Articles

Since 1971, there have been more than 450 published articles regarding Shwachman Diamond Syndrome—which is very exciting! Even though we are involved with such a rare disease, there are researchers who have been working on it for a very long time! It is important to support this research community.



You may view the articles from [a pre-populated article search on the PubMed website](#). PubMed is a part of the U.S. Library of National Medicine, National Institutes of Health. Some of the articles on the PubMed website are only an abstract; however, some have an option to view “free full-text article”.

FAMILY SUPPORT

Part of the Shwachman Diamond Syndrome Foundation's mission is to support patients and families. We strive to link SDS families through the internet and medical family conferences. We also provide families with the most recent medical information, share experiences and provide emotional support. Please use the following support outlets as a way to stay connected with the SDSF and other SDS families. Also, visit www.shwachman-diamond.org for more information regarding medical conferences (Camp Sunshine), the SDS registry, research articles, fundraising, stories about other SDS families, and resources/links available for your benefit.

Call/Email the Shwachman Diamond Syndrome Foundation

The Shwachman Diamond Syndrome Foundation encourages families to connect with us and learn more about how we can assist you. Whether you are newly diagnosed or have been dealing with SDS for a long time, please contact us at 1-888-825-SDSF (7373) or email us at info@shwachman-diamond.org.

- Facebook ([SDSF facebook page](#) and the [private support group](#))
- Instagram - [sdsfoundation](#)
- Twitter – [cureforafuture](#)

INTERNATIONAL SDS SUPPORT GROUPS

AUSTRALIA

Shwachman-Diamond Syndrome Support
Contact: Joan Buchanan
Phone: +61 2 9295 8359
Email: buchanan.joan@gmail.com

CANADA

Mailing Address:
2152 Gatley Road
Mississauga, ON
Canada. L5H 3L9

Toll Free: 1-(866)-462-8907
Email: info@shwachman.org
Website: www.shwachman.org

GERMANY

Contact: Christiane Weyer
E-mail: jane.weyer@web.de
Website: www.shwachman.de
Phone: 05132/589581

ITALY

The Association for Shwachman Syndrome
Contact: Aurelio Lococo
Email: aiss@shwachman.it
Website: www.shwachman.it

NEDERLAND

Shwachman Syndrome Support Group
Email: info@shwachman.n1
Website: www.shwachman.nl

UK

Shwachman Diamond UK
Contact: Kathryn Doddridge
Email: family_matters@shwachman-diamond-uk.org
Website: <http://www.shwachman-diamond-uk.org>

FAMILY SUPPORT (cont.)

Camp Sunshine (<https://www.campsunshine.org>)

SDSF and other SDS families attend Camp Sunshine in Casco, Maine every two years. Attending Camp Sunshine is a great opportunity to meet other families and share your SDS journey with them. See old friends, make new ones and talk to medical experts while your children are cared for and entertained. This is a FABULOUS week for the entire family! SDSF encourages SDS families to attend Camp Sunshine. The only cost to SDS families is transportation to and from Camp.



More information about Camp Sunshine (from the Camp Sunshine website)

We inspire hope in families affected by life-threatening childhood illness through our unique, supportive program at our beautiful campus on Maine's Sebago Lake.

When a child faces serious illness, the entire family is impacted. Camp Sunshine's mission is to provide respite and support to each family member - children, siblings, and parents. Since its inception, Camp Sunshine has offered comfort, hope, and support to over 43,500 individuals from 48 states and 23 countries. Camp Sunshine is currently the only full-time facility in the nation whose sole purpose is to provide respite for the whole family.

The families who come to Camp Sunshine have children diagnosed with cancer, hematologic conditions, renal disease, systemic lupus erythematosus, and who have undergone solid organ transplantation. They attend illness-specific sessions of varying lengths where they have the opportunity to solidify and renew their relationships while meeting others facing similar challenges. Bereavement programming is also provided for families who have lost a child to the illnesses served at Camp.

Recreational activities allow children and adults to relax and enjoy the simplicity and beauty of life along the pristine shores of Sebago Lake. Games and other events foster family involvement, encouraging joy and laughter. While at Camp Sunshine, each family stays in their own suite, equipped with two twin beds, bunk beds, a futon, full bathroom, microwave oven, and refrigerator. Mealtimes in our dining room are occasions for socializing and unwinding.

The Camp Sunshine Program is free

All families are sponsored by generous individuals, civic groups, corporations, and foundations. To learn more about sponsoring a family and other ways of supporting Camp Sunshine, please visit the giving section of this website.

Medical & Psychological Support

Onsite professionals are available to support the medical and psychosocial needs of families attending Camp. A physician is available 24-hours a day throughout each session. In addition, a state-of-the-art children's hospital is only 40 minutes from Camp, and an urgent care center and community hospital with fully staffed emergency rooms are 20 minutes away.

FAMILY SUPPORT (cont.)

With a diagnosis of Shwachman Diamond Syndrome, there are several important documents and records that should be organized to keep track of a patient's care. A well-organized system for storing and filing these documents, whether hard-copy or electronic, is important for the long-term care of the patient.

A binder for organizing patient care

SDSF suggests creating a binder (or an electronic system) to manage your SDS care. In this binder, important medical paperwork should be included such as the patient information sheet, medical team contact information, a log of appointments, a list of medications, a monthly calendar to mark appointments/therapy sessions, a log of lab results, bone marrow biopsy procedures, x-rays and CT scans, and any other pertinent information to meet your specific needs. We recommend purchasing a heavy 3-ring binder and tabs/folders to organize your binder.

We have provided some forms that you can print out to get you started in creating your own care binder. Please visit our website at www.shwachman-diamond.org and look for "Organizational Guide to SDS Patient Care" as a download under the Resources tab.

Electronic tools to help organize patient care

Since technology has allowed us to go paperless in many ways, it may be your personal preference to keep a "digital binder" or to utilize a different digital system to organize your SDS care. The following is a list of suggestions on how to utilize a digital organizational system.

- MyChart
 - This app gives patients (or caregivers of minors) access to a secure, convenient and free way to manage their personal health care information at participating hospitals. Check with your doctor if a patient portal similar to MyChart is available and for the instructions to get set up.
- FollowMyHealth
 - This app gives patients (or caregivers of minors) access to a secure, convenient and free way to manage their personal health care information at participating hospitals. Check with your doctor if a patient portal similar to FollowMyHealth is available and for the instructions to get set up.
- Microsoft HealthVault
 - Microsoft HealthVault helps you gather, store, use, and share health information for you and your family. You can keep all of your health records in one place that's organized and available to you online.
- Evernote
 - This app is great for keeping notes about doctor appointments, keep a list of questions for an upcoming appointment, scan in paper copies of CBC results, doctor's notes, etc.
- Microsoft Excel spreadsheet
 - The binder forms above that can be downloaded have also been compiled into an Excel spreadsheet. Download the spreadsheet, "SDS Patient Care Spreadsheet" on www.shwachman-diamond.org.

Shwachman-Diamond Syndrome Foundation Fundraising Guidelines

Shwachman-Diamond Syndrome Foundation (SDSF) welcomes all efforts by its friends and associates to assist in raising funds to support our mission and related efforts.

As you are aware, SDSF is a tax-exempt organization pursuant to 501(c)(3) of the Internal Revenue Code. In order for SDSF to comply with its By-laws and the Internal Revenue Code and in order for SDSF to keep accurate records, it is necessary that the following fundraising guidelines be followed:

1. To ensure that contributions are tax deductible by the donor, all checks or money orders MUST be payable to SHWACHMAN-DIAMOND SYNDROME FOUNDATION.
2. To host a fundraiser for SDSF, please send an email to info@shwachmandiamond.org so that we can support your event by providing you with bracelets, brochures, the use of our logo, and other supplies as well as helping you advertise your event on our Facebook and Twitter pages.
3. All funds raised in the name of SDSF are expected to be remitted to SDSF.
4. All contributions (including cash) should be directed to the current office of SDSF for recording. The office will then forward contributions to the Treasurer of SDSF for deposit into the proper bank accounts. No other individual is authorized to deposit or endorse checks on behalf of SDSF.
5. Include the name and proper mailing address of each contributor with the contribution. The President is responsible for acknowledging all contributions in accordance with the IRS Code and will keep proof of such acknowledgements with the other financial records of SDSF. In addition to the required notice, anyone is welcome to thank contributors personally.
6. Include any special instructions with the contribution, including but not limited to, the directed use of the contribution (i.e. research, outreach, etc.); the name and address of any individual (other than the contributor) who should be advised of the contribution (i.e. honoree or family of deceased); or whether the contribution should remain anonymous. Contributions may be directed for research; however they may not name specific types of research or research projects.
7. In accordance with its by-laws, SDSF has established bank accounts for the deposit of all funds authorized by the Board of Directors. The signatures on these accounts consist of the current officers of SDSF. No other individuals may open bank accounts on behalf of SDSF or become signatories of any bank accounts without the prior approval of the Board of Directors.
8. All legitimate expenses of fundraising will be reimbursed or advanced by SDSF. For reimbursement, all individuals should submit an itemized list of expenses and copies of receipts. For advances, all individuals should submit an itemized proposal of expenses prior to the event and submit copies of receipts within 30 days of the event along with any excess advance. The Board of Directors reserves the right to disallow any expenses. If you have a question about a specific expense, please contact the Board of Directors prior to incurring the expense.
9. No individual or group shall be authorized to reimburse itself out of funds raised on behalf of SDSF.
10. For reimbursement by SDSF, all individuals must evidence of proper permits, if your state requires them. It will be your responsibility to research the legality of your event in your state and provide evidence of same to SDSF.
11. These guidelines have been set forth for your protection as well as the protection of SDSF. SDSF will not be responsible for any fundraising activity that does not comply with these guidelines and cannot guarantee that any resulting contributions will be tax deductible. In the event that you have any questions regarding these guidelines, please contact the Board of Directors for review of your fundraising activity prior to hosting fundraisers for SDSF.

We appreciate your cooperation with these guidelines and your commitment to SDSF.

FUNDRAISING IDEAS

- **5K Run/Walk**
 - Plan a 5K Run/Walk race or a fun run in your area. You can have t-shirts for participants and medals for winners in certain age groups.
- **Raffle/Silent Auction**
 - Ask local businesses to donate items, packages, or gift cards to your event. Plan a dinner, make tickets, and do a raffle to raise more money.
- **Bounce House Fundraiser**
 - Ask a bounce house business or rent your own bounce house and plan a kid-friendly event. You could also have a silent auction or attendance prizes donated by businesses.
- **Tournament (Golf, Softball, Bowling, Chess, etc.)**
 - Gather friends and family and make some teams to compete in a tournament. You can have t-shirts made and trophies for the winning team.
- **Car Wash**
 - Plan a car wash at a local business that will provide the location and water hookup. Ask friends and family to help wash cars and don't forget to make flyers and signs to advertise.
- **Garage sale/Bake Sale**
 - Plan a garage sale and/or bake sale. Ask friends and family to go through their closets for unwanted stuff that they can donate to your garage sale.
- **T-shirts**
 - Design a custom t-shirt for the patient and proceeds of the shirt sales are donated to SDSF
- **"Direct Sales Company" Fundraiser**
 - Find an independent consultant for a company such as Thirty-One Gifts, Scentsy, Mary Kay, Tupperware, Avon, Premier Jewelry, Pampered Chef, etc. Ask the consultant to donate proceeds to SDSF.
- **Birthday Party**
 - Instead of gifts for your child, you can ask guests to make a contribution to SDSF in lieu of gifts.
- **Dance Party**
 - One family was able to secure a band, get a location donated for their use. They sent out invitations to all their friends telling them that the dance will be a fundraiser for SDSF
- **Carnival**
 - One family was given the use of a park, got donations of carnival games, prizes, food, etc. and had a carnival.
- **Jeans /Free Dress Day**
 - Ask your boss/principal to sponsor a dress-down day at work or school. Each employee or student who participates can pay a fee to be used as a donation.

FUNDRAISING TIPS/TOOLS

- **Six Week Event Planner Guide:**
 - **6 weeks out: Determine what event you think will work best for you and Ask for Help!** Ask your family, friends and colleagues to volunteer to help. You'll want 2-3 people minimum as your organizing committee. Do you want to host a Dance? Party with raffle prizes? Golf or Tennis or Chess Tournament?
 - **5 weeks out: Get Organized!** Set a location, date & time. Determine a fundraising goal, set up a Facebook page or event and invitation, make flyers or invitations. Ask local businesses to sponsor/donate to your event. If you can't get dollars, get in-kind donations such as food, prizes, hall rental. Order SDS rubber bracelets or create and order t-shirts from your local supplier.
 - **4 weeks out: Spread the Word!** Email/mail/share your invitation or flyer. Ask friends and family to volunteer to help at the event. Reach out to civic & religious organizations you are involved with. Contact news media.
 - **3 weeks out: Check Final Details!** Print signs and other materials for event location. Gather donations of refreshments. Stay in touch with your volunteers.
 - **2 weeks out: The Home Stretch!** Send reminders to registrants and volunteers. Double check supplies and materials.
 - **1 week out: Countdown to Success!** Check weather reports. Verify contingency plans. Confirm volunteer assignments.
- **Spreading the Word/Publicity**
 - To get the word out about your event and to help with registration, create a facebook page or event. Have friends/family share the event on their facebook page.
 - Contact your local paper to alert them of the event. This way, you gain more donors and awareness of SDS. Contact the editors of the newspapers or contact a radio/TV station. Make sure they print the full name of the disease and give them a brochure or the web site address for them to get their information from. Also, make sure they print the web site name: www.shwachman-diamond.org.
- **Other Tips/Information**
 - Consider printing a program for your event that acknowledges all donors and pass it out at the event
 - Make sure that you send thank-you letters to all donors for their contributions, whether financial or in-kind items. Please send a list to the Board of any donor who has given over \$250.00 so that they may receive a legal tax deduction letter. You may notify the Board if you want those names to appear in the newsletter on the donor page or if you would like to keep those names privately held. If you ask for privacy, the Board will certainly honor your request. SDSF will send all donors thank you letters.
 - Federal tax disclosure requirements: All charities are required by federal law to let their participants know what part of their entry fee/ticket price is tax deductible and must be printed on the invitation. For example, if you serve dinner with your event valued at \$25.00, and you charge \$75.00 to come to the event, then the deduction is anything over \$50.00. Your disclosure would read: Tax deductible in excess over \$50.00.
 - Do you need SDSF brochures to hand out at the event? Email us at info@shwachman-diamond.org

Shwachman-Diamond Syndrome Foundation

IDEAS FOR A FUNDRAISING LETTER

- 1. Start with a very personal statement.** A plea for help, and a brief description of your child's situation. Some examples follow:

- A. "This is a letter I hoped I would never have to write and I am sure you would never want to receive. However, I must! I hope you will read it and do whatever you can to help.

We have a wonderful and loving grandson, (insert name), now (insert age). He is afflicted with a rare disease called Shwachman-Diamond Syndrome. Words can never express our sadness and devastation..."

- B. "We would like to thank you for your support since our son (insert name) was diagnosed with Shwachman-Diamond Syndrome three years ago.

A few weeks ago, a friend in California asked how her/his health was. He then said, "I feel so helpless. I wish there was something I could do." We told him he can help and make a big difference – not only for our child, but to the many others who suffer from Shwachman-Diamond Syndrome."

- C. "As you probably know, our beloved child (insert name) is in need of a bone marrow transplant to correct immediate life-threatening complications from Shwachman-Diamond Syndrome. It is difficult for us to describe the horrors of this diagnosis. We have watched in agony and despair the downward course of his/her illness.

Medical scientists cannot say with certainty what the long-term future holds for our child. I hate to ask for help, so I have put this off for some time. But my desperate sense of urgency has overcome my reluctance and I am turning to my friends and family for help..."

- D. "As many of you already know, in April we learned that our three-year-old son/daughter, (insert name) has a rare genetic disease called Shwachman-Diamond Syndrome. While his/her diagnosis gives us new insight into his/her previous medical problems, the news has been devastating..."

- E. "Some time ago you responded generously to our appeal for help. I cannot tell you how grateful we were. I want to report that there has been no spectacular progress and ask for your financial help again..."

2. Describe, briefly, what Shwachman-Diamond Syndrome is:

- A. "Shwachman-Diamond Syndrome is a mysterious and often fatal disease..."
- B. "Shwachman-Diamond Syndrome is an inherited, multi-system disorder which affects the pancreas, bone marrow, skeleton, and may affect other organs..."
- C. "These children can be frequently and severely ill with infections that can be life threatening. Patients with Shwachman-Diamond Syndrome are at a higher risk of developing leukemia, bone marrow failure, and other potentially fatal blood diseases..."

3. Describe the tremendous progress being made in research, our reasons for hope, our need for their financial support.

"Researchers are studying the bone marrow and blood of patients with Shwachman-Diamond Syndrome for early signs of myelodysplastic syndrome and leukemia."

"Ongoing research projects are taking place. Information and patient samples are being recruited to survey and test bone marrow and exocrine pancreatic dysfunction. Genetic studies are also being done for molecular genetic studies geared toward tracking the chromosomal defects in order to achieve improvement in diagnosis and care of patients."

"Funds to support these researchers are in critically short supply. This is why we are asking family and friends for help. If sufficient support is available, researchers are confident that they will make significant progress over a period of 3-5 years. Your contribution is vital to this research."

"But much remains to be done. We need to support research which will prolong lives, while we push for a cure for Shwachman-Diamond Syndrome. These researchers desperately need our help!"

"We are turning to our relatives and friends for help. Can you give \$25, \$50, \$100, \$250, or even \$1,000 for this vital cause?"

"Your check should be made out to Shwachman-Diamond Syndrome Foundation. It can be mailed to us and we will forward it or you can mail it directly to the following address: P. O. Box 6723, Florence, KY 41022."

4. Consider adding more personal information about your child at the end of your letter. Repeat your plea for help:

"Johnny is a bright, happy, well-adjusted eight-year-old. He is currently in the second grade and loves school, video games, and the Cincinnati Reds. We are including his picture so you can see what a spunky kid he is!"

“Susie’s enthusiasm for life touches all who meet her. Her compassion for her friends makes her popular and respected by her peers.”

“Watching Johnny and his sister Stacy, age five, play every day, you would never guess one of them might not have a chance to lead a full and productive life.”

“We have no way to know when this tragic disease will strike Peggy hard. We have no way of knowing how rapidly Peggy’s health will deteriorate once it does strike.”

“We can only hope, pray, and do our share to speed the research efforts as they race to find a cause and a cure.”

“We thank you for your concern and your prayers. There is nothing more important to us than the lives of our children. We know you will agree.”

“We are asking for your help. Please send a contribution today. Your gift could be the difference, not only for Billy, but for many others like him.”

“Our deepest and sincerest thanks.”

5. Always add a P.S.:

P.S. Again thanks for your contribution. We can never adequately express our gratitude for knowing that we can count on your help.

P.S. You may get many requests for help. But please know that this cause will help save lives.

P.S. Please keep the picture of Julie as a reminder, each day, that your contribution could make a real difference in giving her and many others the chance for a bright future.

6. A short, handwritten note at the bottom of your letter is extremely effective. Something like this:

“This cause means so much to us-Please help if you can.”

GLOSSARY

For Shwachman-Diamond Syndrome

Updated 2016 by J. Mowery & J. Rommens

GENETICS

Chromosome- structure in the nucleus of the cell which contains the genes responsible for heredity. Normal human cells contain twenty-three pairs of chromosomes. One of each pair is inherited separately from a person's mother and fathers

Gene- heredity unit. Each gene carries the genetic code, to the blue print for a specific protein. Each human cell has about 28,000 genes. About 10-12,000 genes are active in any given cell type.

Gene with mutation- a faulty gene, which may result in an inherited disease.

Recessive- a mutation is said to be recessive if an individual inherits two copies of the gene, one from each parent, to develop an inherited disease. Individuals with one mutation and a normal gene will have no clinical symptoms. They are called "carriers".

SBDS- Shwachman-Bodian-Diamond is the name of the faulty gene that leads to most cases of SDS. The protein product from SBDS is thought to help with the manufacture of proteins from the genes that are active in cells.

BLOOD

Hematology- the study of the blood, blood forming cells and the disorders associated with them.

Bone marrow- soft tissue within the bones where all the different blood cells are manufactured.

Stem cells- cells in the marrow that grow and divide to make more stem cells as well as progenitor cells that lead to the platelets, red cells and white cells.

Plasma- a yellowish fluid in the bloodstream which contains various proteins and other substances. Red cells, white cells and platelets are suspended in plasma.

Hematopoiesis- the production of red cells, white cells and platelets by the stem cells in the bone marrow.

Complete blood count (CBC)- number or percent of blood cells, which include white cells, red cells and platelets.

Red blood cells count (RBC)- small round cells that contain hemoglobin. Hemoglobin, which are necessary to carry oxygen throughout the body. Red blood cells account for almost half of your blood volume.

White blood cell count (WBC)- this test measures the number of white blood cells in the blood. WBC's help fight infections. They are also called leukocytes. The most common kinds of white blood cells are lymphocytes and neutrophils.

Anemia- a decrease in red cells which contain hemoglobin. Hemoglobin, which contains iron, is necessary for carrying oxygen to the body's cells.

Hemoglobin (HGB)- the oxygen carrying pigment of the red blood cells; binds with oxygen in the lungs and is carried to the body's cells.

Hematocrit (HCT)- ratio of red blood cells to plasma in the blood, the portion of the blood's total volume that is made up of red blood cells.

Hemorrhage- bleeding from any site in the body.

Differential count- this is the percent of different types of white blood cells in the body.

Leukopenia- low total white blood cell count.

Neutrophils- (also known as granulocytes)- a mature white blood cell that fights bacteria infection.

Neutropenia- a low absolute neutrophil count (ANC). When the count falls below 1500 per micro liter of blood.

Bands- immature neutrophils. These are usually counted as neutrophils when determining total neutrophils in the blood.

Absolute neutrophil count (ANC)- this is determined by adding the percentage of neutrophils or, polyps in the blood with the percentage of bands in the blood, then multiplying that number by the white blood cells count and multiplying the product by 10. This number represents the number of neutrophils which are available for defending the body against infection.

Cyclic or intermittent neutropenia- when the neutrophil count fluctuates between a normal and a low count. The timing cycle averages about every 21 days, and lasts from 3 to 5 days but can vary.

Severe chronic neutropenia- when the ANC (absolute neutrophil count) is below 500.

Phagocytosis- engulfment and destruction of bacteria or damaged cells by some type of white blood cells, including neutrophils.

Chemotaxis (also known as neutrophil mobility) movement of neutrophils towards a bacterium or an area of tissue damage. Neutrophil must be able to migrate to the particular part of the body to fight off infections.

B-cells- lymphocytes that produce antibodies which help fight infection.

Lymphocytes (T cells or B cells)- cells of the immune system, critical for fighting disease and to help eliminate damaged cells.

Platelets (also known as Thrombocytes)- blood cells that prevent bleeding and bruising. Normal counts range from 150,000 to 400,000. They help to form blood clots.

Thrombocytopenia- a condition in which the number of platelets is less than 100,000 per microliter of blood.

Platelet aggregation- the sticking of platelets to each other to form a clot. This ability can be evaluated by laboratory testing. Abnormal results reflect an increased tendency to bleed (poor clotting), despite a normal platelet count.

Petechia- tiny red dots on the skin due to bleeding under the skin. Usually caused by low platelet counts.

Aplastic Anemia- a rare but extremely serious condition that results from the unexplained failure of the bone marrow to produce blood cells.

Myelodysplastic Syndrome- an abnormal development of the cells produced in the bone marrow. This might be an abnormality in shape, size or organization of adult cells. Abnormal proliferation of cells can progress to leukemia.

Leukemia- cancer of the white blood cells with uncontrolled increase in white blood cell count

Sepsis- an infection of the blood stream or body tissues. Sepsis can be very serious and should be treated immediately.

Blood cultures- these are used to detect the presence of bacteria or fungi in the blood, to identify the type present and guide treatment.

BONE MARROW

Bone Marrow- soft tissue within the bones where all the different blood cells are manufactured.

Stroma- The supporting tissue of the bone marrow.

Bone Marrow Aspiration- test in which a liquid sample of the bone marrow is removed by needle aspiration and examined or tested. Results from this test show the appearance of blood cell precursors. Cytogenetic analysis and other special tests require this type of liquid sample.

Bone Marrow Biopsy (BMB)- A test in which a solid core of bone marrow is removed with a biopsy needle. The biopsy is helpful in determining the cellularity of the marrow, among other things.

Cytogenetics- is a branch of genetics that is concerned with the study of the structure and function of the cells, especially the chromosomes.

Cellularity- in the bone marrow, the degree, quality or condition of cells that are present. Evaluation of cellularity aids in determination of whether cells lines are increased or decreased within the marrow.

Colony Stimulating Factor and other Factors (also known as a hematopoietic growth factors or cytokines)- substances produced by the body, which stimulate the production of certain blood cells. Some of these substances have been manufactured synthetically. Examples are granulocyte stimulating factor (G-CSF or neupogen) and various interleukins.

Graft versus Host Disease (GVHD)- the donated bone marrow or peripheral blood stem cells view the recipient's body as foreign, and the donated cells/bone marrow attack the body.

Bone Marrow Transplant- a procedure to replace damaged or destroyed bone marrow with healthy bone marrow stem cells.

Telomere - is a region of repetitive nucleotide sequences at each end of a chromosome, which protects the end of the chromosome from deterioration or from fusion with neighboring chromosomes.

Blast cells- Immature cells found in the bone marrow. They are not fully developed and do not carry out any particular identity.

PANCREAS

Pancreas- A large gland that lies behind the stomach. It is made up of two parts, the endocrine and exocrine components.

Endocrine- this component of the pancreas secretes insulin and other hormones, which are necessary to control blood sugar.

Diabetes Mellitis- this occurs when the endocrine portion of the pancreas produces none or insufficient insulin. Insulin is a hormone responsible for controlling blood levels of glucose and for the absorption of glucose in the cells of the body. Glucose is needed for energy.

Hypoglycemia- a condition characterized by abnormally low blood glucose (blood sugar) levels, usually less than 79 mg/dl.

Exocrine- The component of the pancreas that produces several digestive enzymes, including amylase, lipase, protease and trypsin.

Amylase- The digestive pancreatic enzyme that breaks down ingested starches.

Lipase- The digestive pancreatic enzyme that breaks down digested fats.

Proteases- The pancreatic enzyme (multiple types) that breaks down digested proteins.

Trypsin- and important pancreatic enzyme which breaks down protein.

Malabsorption- when the body does not absorb nutrients, vitamins and minerals properly. Malabsorption may impair normal growth and development. Failure of the exocrine pancreas or pancreatic insufficiency may cause malabsorption.

Pancreatic dysfunction or exocrine pancreatic dysfunction- evidence of loss of digestive enzyme production in the pancreas.

Pancreatic insufficiency- The pancreas does not produce enough enzymes to digest food.

Pancreatic sufficiency- The pancreas is not normal, low function, but has sufficient function for digestion without the continuing need for pancreatic enzymes with meals.

Steatorrhea- presence of excessive fat in the stool.

Medium chain triglycerides- fats which are most easily absorbed by the body. These can be found in palm oil and coconut oil, for example.

Polyunsaturated fats- fats that are liquid at room temperature.

Fat soluble vitamins- vitamins A, D, E and K; the vitamins that depend on normal digestion to be absorbed properly.

SKELETON

Coxa Vera- a deformity of the hip in which the angle between the neck and the head of the femur, and a shift of the femur (thigh bone) is reduced causing shortening of the leg and a limp.

Genu varus- bowing of the legs at the knee joint.

Genu valgum- knock-knees. The opposite of Genu varus.

Growth plate- this is the hyaline cartilage plate in the metaphysis at each end of the long bone. The plate is found in children and adolescents; in adults, who have stopped growing, the plate is replaced by an epiphyseal line.

Metaphysis- growth plates at the end of the long bones.

Syndactyly- defect in which two or more fingers or toes are joining together.

Thorax- chest.

Tibia- bone of the lower leg, also called shin.

Dysplasia- any abnormality of growth, abnormal size or shape. In Shwachman Diamond syndrome patients, this term may be used to describe abnormalities of the skeleton.

DEXA Scan- dual energy X-ray absorptiometry- a type of one scan that measures bone mineral density.

GROWTH AND NUTRITION

Failure to Thrive- growth failure in an infant or small child. Usually due to inadequate food intake or excessive losses due to malabsorption.

Growth velocity- the rate at which a child grows.

Short Stature- below average height for age, i.e. short, but not malnourished.

Malnutrition- poor nutritional state due to inadequate food intake, a chronic disease or malabsorption. Usually means being underweight.

GENERAL

Bacteria- microorganisms; some can be healthy, such as those that co-habitats and live in your intestine, but they and others can cause serious infections when access other parts of your body, such as blood or kidneys.

Chronic Illness- any long-standing loss or abnormality of bodily function (refers to changes in an individual's body).

Cirrhosis of the liver- extensive scar tissue that forms as the result of damage to the liver, and may lead to decreased liver function.

Endocardial fibrosis- damage to the lining of the heart and valves.

Hepatomegaly- enlarged liver, without necessarily affecting function.

Hypotonia- Poor muscle tone.

Ichthyosis- A condition in which the skin is dry, rough and scaly.

Immunodeficiency- reduced ability of the body's immune system to fight infections.

Intravenous immunoglobulin (IVIG)- is a plasma product used in the treatment of certain conditions related to the immune system.

Intravenous- injection directly into the vein.

Subcutaneous injection- an injection administered into the subcutis, the layer of skin directly below the dermis and epidermis.

Renal tubular dysfunction- a malfunction of the fine tubular part of the kidney, through which water and certain substances are reabsorbed back into the blood.

Xerophthalmia- a disorder caused by vitamin a deficiency that can result in severe damage to the cornea.

Fatty Liver Disease (NAFLD)- is the build up of extra fat in the liver cells that is not caused by alcohol. It is seen when more than 5 to 10% of the liver's weight is fat, it is called fatty liver (steatosis).

Antibody titer- the antibody titer is used to determine whether a previous vaccine helped your immune system protect you against a specific disease.