

## SARDs: What are they?

*In this issue of JointHealth<sup>TM</sup> monthly, Arthritis Consumer Experts introduces you to the little known rare forms of arthritis. Though star tennis player Venus Williams brought one form to the limelight when she was diagnosed with Sjögren's syndrome last year, they remain under-diagnosed and not well understood.*

You may have heard of osteoarthritis and rheumatoid arthritis since there is a good chance you or someone you know lives with one of the two diseases. But have you heard of lupus, Sjögren's syndrome, scleroderma, polymyositis, dermatomyositis, or systemic vasculitides? They are six rare arthritis types that are grouped together because they share symptoms, treatment, and complications. Collectively, they are called **systemic autoimmune rheumatic diseases (SARDs)**.

Individually, with few people having any one of these disease types, research has been difficult and limited. However, studying them together better represents the disease population, making research easier and leading to better quality information. Now, there is a greater understanding of the risks and health complications linked to these diseases. Also, we know the number of people

living with SARDs and the associated healthcare costs, including those caused by hospitalizations, outpatient and doctor visits, and medications.

Having many features in common, all SARDs are chronic autoimmune disorders that lead to inflammation and organ damage. Inflammation is caused by the body's immune system attacking and destroying its own tissue. Depending on the specific disease type, white blood cells can invade muscles, skin, internal organs, arteries and blood vessels. Being systemic diseases, they can affect the whole body.

The complications they share include cardiovascular (heart) disease, osteoporotic fractures, kidney failure, infections, and cancer. Except for vasculitis, SARDs mostly affect women. They can occur at any age, though each type has a window during which it will most likely arise. Sometimes other conditions, such as Raynaud's phenomenon (blue hands with cold), can go along with the SARDs. There is no cure for any of these diseases but they can be controlled, especially when they are diagnosed early.

There is some evidence to suggest the diseases are genetic (inherited), but which genes and how they are turned on or what role they play is not well understood. In some types, it is possible that a viral infection could trigger disease. Because SARDs more typically affect women, it is thought hormones may be involved. Researchers still have a lot to learn before they will know for certain what causes these diseases. Studying them together is getting them closer.



In British Columbia alone, there are 18,741 people living with a form of SARD (about 0.5% of the adult population).

### The BC cumulative total healthcare costs for SARDs over a 12 year period

 Hospitalization costs	\$291,664,950
 Out patients and doctor visits	\$154,580,563
 Medications	\$124,971,267
 Total	<u>\$571,216,779</u>

Canadians living with SARDs face significant challenges getting a timely diagnosis and accessing appropriate treatment, leading to more doctor visits, increased hospital stays and higher medication costs to the healthcare system.

## About lupus

Systemic lupus erythematosus (SLE) affects about one in 1000 Canadians, usually striking between the ages of 15 and 45. For every nine women with lupus, one man will have the disease.

Tissues affected by lupus can include the skin, joints, muscles, kidneys, lungs, heart, blood vessels, and the brain. Because lupus can affect so many different areas of the body, it often presents very differently among patients, making it difficult to diagnose.

Warning signs include:

- Swollen or stiff and painful joints
- Unexplained fever
- Loss of appetite and weight loss
- Fatigue and low energy
- Skin rash, especially if brought on by sun exposure
- Hair loss (on the pillow)
- Ulcers in the mouth and/or nose, usually painless
- Pain in the chest while lying down or taking deep breaths
- Low blood counts, including anemia
- Clots in young individuals (legs, lungs or brain)
- Unexplained recurrent pregnancy losses
- Protein or blood in the urine, seen only under the microscope

While the exact cause or causes of lupus remain unknown, there are a number of factors researchers believe may trigger the disease, either alone or in combination with one another. These include genetics, smoking, hormones, certain types of antibiotics and other medications, prolonged and severe stress, viruses, and sun exposure.

## Raynaud's phenomenon

Raynaud's phenomenon is a condition in which there is an exaggerated blood vessel tightening in response to emotional stress or to the cold, restricting blood flow to certain areas of the body, usually the fingers, but sometimes the toes, ears, or the end of the nose. Affected areas generally first turn white and then may become blue. They become red as they begin to return to normal.

Raynaud's can occur on its own or with other types of arthritis. When it is associated with another disorder (such as lupus, scleroderma, or vasculitis), it is secondary. When it occurs alone, it is referred to as primary Raynaud's phenomenon and does not increase the risk of suffering SARDs.

## About Sjögren's syndrome

About 90% of those diagnosed with Sjögren's syndrome are women. It tends to strike around middle age, usually between the ages of 45 and 55.

In Sjögren's syndrome, moisture-producing glands are attacked by the body's immune system, so the most common early symptoms are dry eyes and mouth. Since the dryness can also be due to aging, hormonal disorders, menopause, and certain medications, Sjögren's syndrome is difficult to diagnose.

Warning signs include:

- Very dry eyes, which may feel "gritty"
- Dry mouth, sometimes making swallowing difficult
- Dry nose and throat
- Dry vagina
- Swollen salivary glands
- Fatigue
- Joint pain, swelling, and stiffness, most often in smaller joints like fingers and toes.

The disease can also affect the joints, muscles, and organs including the liver, pancreas, kidneys, lungs, stomach, and brain.

Half of those diagnosed with Sjögren's syndrome also have another form of arthritis, like lupus, scleroderma, or rheumatoid arthritis. Combined with another disease, it is called secondary Sjögren's syndrome. Primary Sjögren's syndrome is the name for the disease when it occurs on its own.



**Marilyn Muldoon**, with the Sjögren's Society of Canada, writes about her experience of living with Sjögren's on the Arthritis Research Centre of Canada's website.

<http://bit.ly/sjogrens>

## About polymyositis and dermatomyositis

Onset most commonly occurs in one's 30s to 50s and is more likely to strike in women.

Polymyositis causes inflammation of the muscles that support and move the skeleton, especially the hips, thighs, shoulders, upper arms and neck. Symptoms develop slowly over weeks or months and include:

- Symmetrical (happening in the same place on both sides of the body) muscle weakness that worsens over time
- Difficulty swallowing and/or speaking
- Mild joint or muscle tenderness
- Fatigue
- Shortness of breath

Remission (symptom free period) is rare, but muscle strength can be improved by treatment.

When polymyositis comes with a skin rash, it is called dermatomyositis.

Dermatomyositis can also affect children, usually between the ages of 5 to 15 years. In adults, it more often shows up starting in the late 40s until early 60s. Besides the symptoms listed above, when children get the disease they are more likely to have hard deposits of calcium under the skin, GI ulcers and intestinal perforations.



**Joyce Ma**, member of the Consumer Advisory Board, writes about her experience of living with polymyositis, on the Arthritis Research Centre of Canada (ARC) website.

<http://bit.ly/polymyositis>

## About systemic vasculitides

This very rare SARD that mainly affects men is made up of around 20 different disorders. Examples include: Behcet's disease, giant cell arteritis, Takayasu's arteritis (this one usually occurs in young women), Wegener's granulomatosis, polyarteritis nodosa, and polymyalgia rheumatica. Age of onset depends on the form of vasculitis, though as with most types of arthritis, it can occur at any time.

Depending on the form of vasculitis, it can affect small, medium, or large blood vessels, so that any organ system can be involved: skin, joints, lungs, kidneys, gastrointestinal tract, blood, sinuses, eyes, nerves, and the brain. Therefore symptoms vary widely. They include:

- Unexplained and recurrent fever
- Fatigue
- Weight loss
- Muscle and joint pain
- Loss of appetite
- Nerve problems (numbness or weakness)
- High blood pressure in young individuals
- Blood and protein in the urine
- Pulses hard to find or absent

Sometimes vasculitis occurs without any known cause and other times it can be caused by an underlying disease, such as infection, another immune system disease, an allergic reaction to a medication, or a blood cell cancer.

## About scleroderma

Scleroderma affects about 20 people out of 100,000 and occurs in about four times as many women than men. It tends to develop most often between the ages of 45 and 65.

Scleroderma—the word means "hard skin"—causes the body to respond to an attack from its immune system by producing too much collagen. This can happen anywhere on the skin and on

joints. There are two main types of the disease: systemic and localized scleroderma. The systemic form is more serious. As it progresses, symptoms and complications may include muscle weakness, digestive system problems, Sjögren's syndrome, reduced dental health, kidney and lung problems.

Joint pain and stiffness, particularly in the hand joints, is almost always present at disease onset. Raynaud's phenomenon is also a very common first symptom of scleroderma and is thought to occur in 98% of cases.

Besides the usual suspected causes, some researchers believe that frequent exposure to solvents and heavy metals—for example, due to working in manufacturing or refining plants—may increase the risk of developing scleroderma.



**Melissa Patton**, board member of The Scleroderma Association of BC, writes about her experience of living with scleroderma, on the Arthritis Research Centre of Canada's website.

<http://bit.ly/scleroderma2>

*For a list of arthritis websites, including for SARDs, please visit our resources section at [jointhealth.org/resources-links.cfm](http://jointhealth.org/resources-links.cfm)*

## Pregnancy:

### What are the concerns?

With any of the SARDs, if the disease is active, pregnancy can worsen the symptoms and put the baby at risk. The good news is that when the diseases are well-managed and in remission for at least 6 months, any pregnancy or birth risks associated with them are very small. If you wish to get pregnant, it is important to inform your physician or rheumatologist so they can monitor you and your baby. Also, they may need to adjust your medications, since certain types should not be taken during pregnancy and while breastfeeding. Use of hydroxychloroquine, a common medication used in SARDs, is safe during pregnancy and lactation.

In rare cases, women with lupus or Sjögren's syndrome may pass autoantibodies to their baby, so he or she may be born with a mild malar rash (the characteristic butterfly shaped rash associated with lupus in adults), but be otherwise healthy. Called neonatal lupus, the condition usually disappears by the time the baby reaches six months. In very rare cases, the infant may have developed congenital heart block in utero (in the womb) and need surgery as soon as they are born.

## Q and A with Dr. Antonio Aviña-Zubieta, Research Scientist, Arthritis Research Centre of Canada



### What makes SARDs difficult to diagnose?

**A:** SARDs are difficult to diagnose because they are rare and so most currently practicing physicians did not receive the training to diagnose them. Currently, SARDs are included in medical training so younger doctors are more familiar with SARDs; however, they only see the diseases about once per year, so they do not necessarily have the skills and experience to consider SARDs and make a diagnosis. Furthermore, symptoms vary so much that they are not usually the first to be considered.

Because the diseases look so different among patients, their individual symptoms could fit any number of diagnoses. For example, an unexplainable constant fever could be thought to be a viral infection or cancer. Another example is when a younger person has a stroke. The doctor may not realize it could be caused by a SARD, such as SLE, so may not ask for the right tests to find out. Other times, a young individual may have blood in their urine or high blood pressure, but the doctor may never consider that these symptoms could be due an inflammatory disease, and again, they would not ask for the right tests.

Sometimes symptoms develop slowly over time. They may start with a fever, then arthritis may develop, and then finally the kidneys may become inflamed before the patient accumulates enough criteria to lead to a diagnosis of a SARD. It is key that clinicians start considering these other possibilities, but this may take time and training.

Also, patients may have difficulty describing what is happening to their bodies. Or, since fever does not usually require medical treatment, they may take over-the-counter medications instead of going to their doctor.

### Do people with rarer forms of arthritis feel more isolated?

**A:** Yes. They do. Since there are very few cases, it is hard to find, or create, a network that provides resources for people with SARDs. There are a lot more social networks and support groups for

more common diseases like rheumatoid arthritis and osteoarthritis. Because of this, recently diagnosed patients do not know where to go and feel isolated.

Sometimes people are afraid of getting fired from their jobs or being treated differently, or simply do not know what to do, so this may even hold them back from seeking care.

### What advice would you give to someone who is first diagnosed?

**A:** First, do not panic. It is very common to see people get scared when they hear their diagnosis, especially if they have only heard outdated information. For example, five decades ago lupus was fatal (95% diagnosed with the disease died within 5 years). Now research has changed that and outcomes are dramatically better. With proper diagnosis and early intervention, people survive up to 20 years longer in 80% of cases.

Second, get the support you need. Seek help from your family, friends, and healthcare providers. If you do not have any of those resources then ask for a referral, especially if you are experiencing emotional distress. Specialists that are very helpful include psychiatrists and rheumatologists. If a rheumatologist is not available, internists (internal medicine specialists) are also very useful because of their familiarity with SARDs.

Finally . . . seek care. Do not feel embarrassed or ashamed of your disease. Anybody of any age can have it, so don't be afraid to get help. Learn as much as you can about your disease. Rely on good sources that are not sponsored by companies that sell cures. And, be aware that early diagnosis improves outcomes, so take action.

*Interview adapted from the JointHealth™ workshop titled "Rare Forms of Arthritis". To watch the video, please visit [jointhealth.org](http://jointhealth.org)*

# About Arthritis Consumer Experts

## Who we are

Arthritis Consumer Experts (ACE) provides research-based education, advocacy training, advocacy leadership and information to Canadians with arthritis. We help empower people living with all forms of arthritis to take control of their disease and to take action in healthcare and research decision making. ACE activities are guided by its members and led by people with arthritis, leading medical professionals and the ACE Advisory Board. To learn more about ACE, visit [www.jointhehealth.org](http://www.jointhehealth.org)

## Guiding Principles

Healthcare is a human right. Those in healthcare, especially those who stand to gain from the ill health of others, have a moral responsibility to examine what they do, its long-term consequences and to ensure that all may benefit. The support of this should be shared by government, citizens, and non-profit and for-profit organizations. This is not only equitable, but is the best means to balance the influence of any specific constituency and a practical necessity. Any profit from our activities is re-invested in our core programs for Canadians with arthritis.

To completely insulate the agenda, the activities, and the judgments of our organization from those of organizations supporting our work, we put forth our abiding principles:

- ACE only requests unrestricted grants from private and public organizations to support its core program.
- ACE employees do not receive equity interest or personal "in-kind" support of any kind from any health-related organization.

- ACE discloses all funding sources in all its activities.
- ACE identifies the source of all materials or documents used.
- ACE develops positions on health policy, products or services in collaboration with arthritis consumers, the academic community and healthcare providers and government free from concern or constraint of other organizations.
- ACE employees do not engage in any personal social activities with supporters.
- ACE does not promote any "brand", product or program on any of its materials or its website, or during any of its educational programs or activities.

## Thanks

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