NATIONAL CENTER FOR CASE STUDY TEACHING IN SCIENCE

A Misdiagnosed Odyssey:

Identifying the Origin of Chronic Joint Instability

by
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Part I – The Orthopedic Specialist

Tahlia Brown was 49 years old when her problems first began while working as a nurse in the obstetrics department of a community hospital in California. Overall, her health was good, however she started to notice that her skin was bruising easily, and it seemed somewhat fragile. Tahlia regularly went to the gym, but unfortunately during her exercise routine she injured her lower back, and her physician ordered an MRI. The results revealed some joint instability in the lumbar region, and her doctor referred her to an orthopedic specialist, Dr. Ross. After a year of physical therapy Tahlia expressed concern to Dr. Ross that the pain was not going away, and he recommended that she make an appointment to schedule an epidural. Dr. Ross explained that an epidural injection is a procedure that involves injecting steroid medication into the epidural space around the spinal cord. He told her to think of the epidural space as a sleeve around the spinal cord that gives rise to the roots of the spinal nerves, and that the injection would numb the nerves and decrease inflammation. Tahlia had the epidural steroid injection, and although she initially received some relief, within a year she was having back pain again. Based on her chronic back pain, Dr. Ross recommended another epidural, and after that she felt better.

A year later Tahlia suffered from constant pain in her neck. Once again, she had physical therapy and was given pain medication for six days, but intense pain persisted and eventually her doctor ordered an MRI of her neck. Based on the MRI results, she was referred to a surgeon. The surgeon diagnosed Tahlia with degenerative disc disease and recommended that she have an anterior cervical fusion between two cervical vertebrae. Dr. Ross explained that the surgery was called an anterior cervical discectomy, a procedure in which the disc between two cervical vertebrae is removed to relieve pressure on the nerve root. Once the disc is removed, a piece of bone is inserted in place of the disc, and as the bone grows the vertebrae fuse together. Dr. Ross explained that this procedue would decrease the instability in her neck and reduce the pain. Tahlia agreed to the procedure, and after recovering from the surgery, she felt good enough to return to work delivering babies.

Questions

1. Describe the structure and function of the intervertebral joints in the cervical and lumbar regions. What structures pass through the vertebral foramen and the intervertebral foramen?

2. Why would physical therapy be indicated for lumbar vertebral instability?

3. How did the steroid epidural injection control Tahlia's pain?

4. Following a cervical discectomy, how can the bones fuse together and what impact will that have on stability and range of motion to Tahlia's neck?

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Part II – The Rheumatologist

A year after returning to work at the hospital, Tahlia began experiencing episodes of tachycardia and dizziness. Tahlia attributed this to the stress of her job, however after a few days Tahlia started to have additional joint pain mostly on her left side. At this point in time most of the pain was to her left wrist, which was a big problem since she was left-handed. In addition to her wrist, her lower back pain started again, and she began to have pain in her left hip, shoulder, and ankle. Based on all these developing symptoms with her joints Tahlia's doctor referred her to a rheumatologist who ordered blood tests. The tests that he ordered for Tahlia are shown in Table 1.

Table 1. Test results for Tahlia Brown.

Test	*Normal Range*	*Tahlia's Test Results*
Antinuclear antibody titer	Less than 1:40	1:33
Rheumatoid factor	Less than 20 U/mL	15 U/mL
C- reactive protein	< 3 mg/L	1 mg/L
Erythrocyte sedimentation rate	0 –20 mm/hr.	6 mm/hr.

Tahlia was given an MRI of her left wrist and was referred back to her surgeon who suggested that she should have surgery to repair the tendons in her left wrist. She had the surgery, but recovery was very slow and eventually prevented her from returning to work. At this point in time her lower back issues were causing considerable pain again and since several epidural injections were not eliminating her pain on a long-term basis, she had surgery on her lower back. Shortly after the back surgery she started to develop a burning sensation in her hands and eventually in other parts of her body. Tahlia was referred to a neurologist.

Question

1. Why did the doctor order a rheumatoid factor test and an antinuclear antibody test, and what is your interpretation of the results?

Part III – The Neurologist

Dr. Long ordered a skin biopsy, and Tahlia was diagnosed with small nerve fiber neuropathy. The neurologist believed that Tahlia had a connective tissue disorder based on her symptoms that affected so many parts of her body. Genetic testing was recommended and confirmed the doctor's suspicion. The gene mutations identified were *COL5A1* and *COLA52,* which indicates a specific type of Ehlers-Danlos syndrome (EDS). Dr. Long discussed with Tahlia her diagnosis and how it might impact her life in the future. She explained that collagen is the most abundant protein fiber in the body and is an integral part of our skin, ligaments, and joints. In Tahlia's case, this protein was defective and creating damage in various parts of her body. In the future, she explained it could affect additional joints and bring about more symptoms and pain. Once Tahlia was diagnosed appropriately through genetic testing, her treatment centered around pain management, physical therapy, and the use of supportive braces. Her EDS has worsened over the years, and she now needs to use oxygen intermittently. However, she has found that wearing braces on her neck, wrist, and ankles has provided support to her joints, and certain medications have been able to control some of her pain.

Questions

1. What is collagen? Describe its function and where it is found in the human body.

2. Why did Tahlia's condition impact so many of her joints?

3. The genes *COL5A1* and *COLA52* were identified with genetic testing. What type of EDS does Tahlia have and what clinical manifestations are linked to this type of EDS?

4. Why do you think that she developed small nerve neuropathy?

5. Why did it take years for Tahlia's doctors to identify that she had EDS?

6. In Tahlia's case, is EDS an autosomal dominant or autosomal recessive disease? Explain what these different terms mean and construct a Punnett square for EDS diagnosed in Tahlia's case if her parents are heterozygous.

7. How can medication and adaptive supports be utilized to help a patient with EDS?