Neurofibromatosis Patient with Nephrolithiasis

Case Study # 3

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Neurofibromatosis Patient with Nephrolithiasis

Identifying factors

J.C. is a 28 year-old Caucasian male who presented to the Family Health Center of Southern Oklahoma (FHCSO) on 2/05/10. J.C. is an established patient of our clinic. I chose this patient because he presented with what at first appeared as a benign complaint, right costovertebral tenderness and lower lumbar pain, however, after examination, there was suspicion that he might have nephrolithiasis. I had never seen this patient prior to his visit. Basically, the patient came to our clinic due to the sliding fee scale and his continued pain.

Subjective

Chief Complaint

"My back is really hurting. No matter what position I get in I still hurt."

History of Presenting Illness

J.C. presents to the clinic with complaints of right costovertebral tenderness for the past few days. He states the pain occasionally radiates across his lumbar area and that the pain is worse with activity and after lying on his right side. He is unable to lie on his right side due to pain in his back. He states the pain is decreased to a "4" when lying on his left side. He rates the pain a "7" most of the time. J.C. states that over the past few days the pain has become so intense he has become nauseated. He denies fever, dysuria, or hematuria. J.C. denies numbness or tingling in his legs. He has been taking Ibuprofen for the pain, with little effect. He denies injury. J.C. states he usually doesn't have any joint pain or stiffness.

Past medical history

Illnesses:

1. Gastroesophageal Reflux (GERD)

Allergies: NKDA

Surgeries: None

Medications: Omeprazole 20mg (1) every day

Health Maintenance: Td <10 years ago; Flu shot October 2009; Last Eye exam June 2009, Last dental exam > 2 years ago; No pneumonia vaccine

Family history:

JC.'s mother died at age 50 from injuries sustained from a motor vehicle accident and his father died at the age 42 due to a tractor accident. He has one brother, who has hypertension. J.C. is married and has two children: a five year-old daughter and an 18 month- old son. The children are both healthy except the five year- old daughter is presently being worked up at the O.U. Health Science Center for symptoms of a learning disability.

Social History:

He is married and lives with his wife in a small rural home in Southeastern Oklahoma. J.C. is a welder at a manufacturing company. His wife stays at home and cares for their two small children. They have no insurance and do not qualify for Soonercare (our state's Medicaid program). J.C. states they were financially unable to have an insurance premium taken out of his paycheck. He denies smoking, alcohol or recreational drug use. He states he had smoked one pack a day for two years but quit five years ago. He admits that he does not have a regular exercise program. He denies travel any outside the country.

Review of Systems

General: He reports fair energy levels and denies fever, weight gain or loss, and night sweats.

Skin/Hair/ Nails: No excess sweating, dryness, hair loss or nail changes. States he has some light brown spots on his back for several years. States he has several

	"cysts" scattered on his torso. He states he first noticed the lesions when he was a teenager; always felt embarrassment with his shirt off among his friends. He states they are worse on his right lower lumbar area than on his left.
Eyes:	No vision changes, floaters, scotomas, tearing or drainage from the eyes, no diplobia, or loss of vision.
Ears:	Denies hearing loss, no drainage from the ears, or pain.
Nose:	No head congestion, drainage, sinus problems or epitaxis.
Throat:	No sore throat, hoarseness or difficulty swallowing; No mouth pain or ulcerations.
Neck/Lymph:	No lumps or swelling in his neck that he is aware of. No tender lymph nodes.
Chest/Lungs:	Denies dyspnea with exertion. Denies orthopnea, or paroxysmal nocturnal dyspnea (PND). States he sleeps on one pillow. Denies Tb exposure. Denies shortness of breath or cough.
CV:	Denies chest pain, edema or palpitations.
GI:	Appetite good; C/o occasional heartburn; States he only has difficulty with indigestion 1-2 times a week now since on medication. Prior to getting on omeprazole was having daily episodes. Denies constipation or diarrhea. No black or tarry stools.
GU:	Negative for incontinence or dysuria. No Nocturia; Urine stream strong; No hesitancy.
Endocrine:	No temperature intolerance, no c/o polydipsia or polyphagia. No weight loss or gain
M/S:	States for the past few days he has lower right side CVA tenderness. States the pain radiates to lower lumbar region. Today the pain has increased in severity that it is very painful for him to even walk down the hall. Rates pain "7" most of the time but occasionally decreases to a "4". Denies radicular signs or symptoms. No swelling in joints or redness. Denies muscle weakness. Aggravates= walking and usually only lying in one position too long however, today all movement is painful. Alleviates= usually lying down. Describes the pain as "stabbing" when at "7" and "dull" when at a "4".
Extremities:	Denies edema or hair loss; No cyanosis or coldness of extremities.
Hematological:	No excessive bleeding, cuts that won't heal, or bruising easily.
Neuro:	No dizziness, or seizure activity; No weakness or decreased sensation. No tremors. States he has only an occasional headache, approximately one a month. States usually relieved with Tylenol.
Psych:	No feelings of sadness or depression. No nervousness or anxiety.

Objective

Vital Signs: Ht: 5’10 Weight: 194 BMI: 27.8Temp: 98 Pulse: 88 R: 22 B/P: 130/66

General:	J.C. is a 28 year-old Caucasian male in no acute distress. Well developed & well nourished. Appearance is consistent with stated age. He is dressed appropriately for the weather and is well-groomed.
Skin:	His skin is warm, pink and dry with no excessive dryness. Turgor is brisk. Multiple nodular lesions scattered on torso, ten lesions ranging from 3mm to 2cm. across right CVA area. Nine flat dark beige macular lesions (café au-lait spots) noted across lumbar region
Hair:	Normal texture and distribution.
Nails:	No clubbing noted. Nail beds are pink with brisk capillary refill. No nail deformities noted.
Head:	Normocephalic. Temporal arteries are without bruits. No lesions on scalp noted.
Eyes:	Conjunctivae clear; Sclerae white; Corneae clear; No tearing or drainage; Pupils equal, round and reactive to light and accommodation. Extraocular movements intact in all six fields without nystagmus. Fundoscopic Exam: No hemorrhages or exudates noted; No corrective lens are worn.
Ears:	Canals clear without drainage or excessive cerumen; TM's pearly gray with visible landmarks bilaterally;
Nose:	Septum midline; Mucosa pink; no congestion, drainage or sinus tenderness;
Throat:	Oropharynx without erythema or exudate; Tonsils 1+; symmetrical rise of uvula and soft palate; Tongue midline without deviation or fasciculations; No mouth lesions or masses palpable. Few dental caries noted, gums are pink, with minimal swelling.
Neck:	Supple, without masses or lymphadenopathy; Trachea midline; Thyroid smooth, without enlargement or nodules. No difficulty swallowing
Chest/Lungs:	Respirations unlabored. No use of accessory muscles. Thorax is symmetrical. Breath sounds clear bilaterally.
Heart:	Regular rate and rhythm. S1S2 present without murmur. No clicks, rubs or gallops. Distant heart sounds. No heaves or lifts; PMI at 5th ICS along the MCL.
Abdomen:	Soft, non-tender. Bowel sounds + in all 4 quadrants. No rebound or guarding. No hepatosplenomegaly. No masses or lesions noted. No aortic, renal or illiac bruits noted; Positive CVA tenderness noted on right.
GU:	No inguinal or axillary freckling.
Musculoskeletal:	Joints symmetrical, without swelling, redness or tenderness; Full ROM of all other joints. Strength is symmetrical at 5/5.Tenderness noted to vertebral column along lower lumbar area. Good muscle strength, able to maintain flexion against resistance. Tenderness and guarding noted at right CVA region.
Extremities:	Extremities are pink and warm with no cyanosis or clubbing. Peripheral pulses are 2 + bilaterally. There is no edema and no varicosities are noted.

Neuro: CN II-XII grossly intact. DTR's are symmetrical and 2+; He is alert and oriented to person, place, and time. Short and long term memory and concentration intact. Speech is clear and appropriate to subject.

Psych: Pt is cooperative, with normal reactions to surroundings and questions. His facial expressions, conversation and affect are appropriate to the situation. No outbursts or inappropriate conversation.

Diagnostic Tests:

Helical CT ABD with (with stone protocol)

Reference: CT Scan (Computerized Axial Tomography) is a procedure that assists in diagnosing kidney stones, tumors, bony structures, and infections in the organs and tissues of the body (Tierney,McPhee,& Papadakis, 2006). The IVP was formerly the gold standard, but prospective trials have shown that computed tomography is the best method for diagnosing ureteral calculi (Miller & Lingeman,2007).This test was ordered since one of the differentials was to rule out nephrolithiasis and also due to the increased amount of tenderness in the right CVA. The patient was private pay but Mercy Hospital in Ardmore,Okla has a charity program called the Sisters of Charity; we can enlist patients when we feel tests are indicated and the patient has no funds available. The patient was able to have this test done this morning for a cost of $25.00 after the paperwork for the Charity Assistance was completed.

CBC:	Within normal limits
WBC	8.1
RBC	4.01
Hgb	12.6
Hct	40.2

UA:	Within normal limits except hematuria noted
pH:	7.0
Specific gravity:	1.020
Glucose:	Neg
Ketones:	Neg
Blood:	Trace
WBC:	Neg

Reference:
Complete blood count was done to detect infection or anemias. Urinalysis was done to determine if patient had evidence of hematuria, WBC, or ketones. After UA was done which revealed trace blood, along with the patient's continued CVA tenderness, a CT abd with stone protocol was ordered (Seller,2000).

Assessment

Current Diagnoses

1. Gastroesophageal reflux (GERD) (530.81) (Controlled)
   Reference: GERD is backward movement of gastric contents into the esophagus. Three normal defenses prevent reflux from occurring: lower esophageal sphincter incompetent, esophageal clearing and gastric emptying (Seeler, 2000).

New Diagnosis

1. Neurofibromatosis (237.71) (New Diagnosis)

   Reference: Neurofibromatosis is an autosomal dominant genetic disorder in which the tumors grow along the nerves. Skin changes and bone deformities also occur. The disorder is characterized by multiple, light tan colored café-au-lait spots 5mm or larger

Differential Diagnoses:

2. Nephrolithiasis (592.0) (Working Diagnosis)

   Reference: Kidney Stones may cause intense pain in the back, abdomen, groin, or genitals. Kidney stones can be a cause of CVA tenderness, hematuria, abdominal pain, lumbar and groin pain. (Tierney,McPhee,& Papadakis, 2006).

3. Lumbosacral strain (724.5*)*

   Reference: Lumbosacral strain is trauma to the muscles and tendons of the lower back. Many conditions may cause low back pain, including disc herniation, infection, malignancies, fractures, and neurological conditions. There may be paravertebral tenderness, tightness of muscles in the lower back, positive straight leg raise, and decreased range of motion (Domino, 2008).

Assessment of Presenting Complaint

The patient presents with an acute onset of right costovertebral tenderness, lasting for the past few days. He describes the pain as “sharp, stabbing” in his right costoverterbral region with any kind of movement and lying on his right side. The pain has increased in severity with pain now with any kind of movement. Typical symptoms of acute renal colic are

intermittent, colicky flank pain that may radiate to the lower abdomen, and may have accompanying nausea and vomiting (Miller & Lingeman, 2007). The urinalysis showed a trace of blood; therefore, it was my impression that he could have nephrolithiasis. I also knew he showed evidence of neurofibromatosis with multiple neurofibromas and café-au-lait spots noted. The patient had never been told he had this disorder. I also wanted to discuss this further with the patient but due to his increased pain, I will discuss further at his next visit. My plan was to research this disorder to identify if the pain could be attributed to the multiple neurofibromas located in the right CVA region. Some people with neurofibromatosis 1 develop cancerous tumors that grow along nerves (Theos & Korf, 2006). In my research, even though these neurofibromas multiply and in this patient were increased in the area of his pain, the evidence-based literature stated that only in rare cases does the spinal neurofibromas cause nerve root compression, and compress the spinal cord (Theos & Korf, 2006).

Plan with Rationale

I was able to get the patient in that morning for the CT scan and I called the radiologist that afternoon for the results. The CT scan returned positive with a renal caculi size of 11 mm. I consulted with Dr. Diacon, urologist regarding further treatment. Studies by Albala, Assimos, Clayman, Denstedt, & Grasso (2001) have demonstrated that percutaneous nephrolithotomy was more effective than shock wave lithotripsy in the treatment of calculi >1cm. So due to the size of the patient's stone, he will mostly likely have this procedure done. The patient was notified of the results of his CT scan; discussed options of treatment, and his appointment to see Dr. Diacon on Monday. Conservative treatment was started with pain management. I offered J.C. an injection in the office for pain relief and gave prescription for nausea and pain control until his procedure on the February 8.

General Therapeutics

1. Increase oral fluids/ discussed the importance increasing fluids
2. Hydration and rest
3. Admission to hospital due to increased pain; procedure scheduled the next morning
4. Educated on the use of a strainer for urine and if stones are found bring stone in for analysis.

Medications:

1. Toradol 30 mg IM

   *Reference**:** Ketorolac is not a narcotic but it is indicated for acute pain that requires analgesia at the opioid level. The total combined duration of use of Toradol should not exceed 5 days. It works by reducing hormones that cause inflammation and pain in the body. (www.drugs.com). There is evidence proved that ketorolac combined with a narcotic resulted in greater pain reduction than either group alone (Safdar, Degutis, &Landry, (2006).*

2. Nubain 10 mg IM

   Reference: It is a synthetic opioid agonist-antagonist analgesic. Nubain is a potent analgesic. Its analgesic potency is essentially equivalent to that of morphine on a milligram basis.The onset of action of Nubain occurs within 2 to 3 minutes after intravenous administration, and in less than 15 minutes following subcutaneous or intramuscular injection (www.drugs.com). Patient had no nausea at this visit but was having increased pain during our visit. I used the combination of drugs Toradol and Nubain for better pain control.

3. Lortab 5/500 (1) Q 4-6 hrs prn pain

   Reference: Treating moderate to moderately severe pain. Hydrocodone is a semisynthetic narcotic analgesic and antitussive with multiple actions qualitatively similar to those of codeine. Most of these involve the central nervous system and smooth muscle. The precise mechanism of action of hydrocodone and other opiates is not known, although it is believed to relate to the existence of opiate receptors in the central nervous system. In addition to analgesia, narcotics may produce drowsiness, changes in mood and mental clouding (www.drugs.com).This was offered to the patient due to their increased pain until the procedure could be scheduled.

4. Phenergan 25mg (1) Q 4-6 hrs prn nausea

Reference: This medication is prescribed to prevent and control nausea and vomiting and to prevent and treat motion sickness. Promethazine is well absorbed from the gastrointestinal tract. Clinical effects are apparent within 20 minutes after oral administration and generally last four to six hours, although they may persist as long as 12 hours. Promethazine is metabolized by the liver to a variety of compounds; the sulfoxides of promethazine and N-demethylpromethazine are the predominant metabolites appearing in the urine (www.drugs.com).

Continuity of Care

I have not seen the patient since the initial visit but he was seen by another provider at our clinic. I did however, talk to him over the phone. He had to have a nephrolithotomy due to the size of the stone. Dr. Diacon had attempted lithotripsy but this was unsuccessful. He is doing well and his pain is almost resolved. He only experiences "dull pain", rating it a "1". He seldoms takes pain medication now and is doing much better. He states he has returned to full activity. He denies fever, hematuria, or dysuria. We discussed the diagnosis of neurofibromatosis and I was able answer several of his questions. Patient states he is not able to return for follow up until the end of the month but I assured him if he needs anything or has questions to call our clinic and I would be glad to assist. He was appreciative of the help that was given.

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