PedsCases Podcast Scripts

This is a text version of a podcast from Pedscases.com on "Juvenile Dermatomyositis." These podcasts are designed to give medical students an overview of key topics in pediatrics. The audio versions are accessible on iTunes or at www.pedcases.com/podcasts.

Juvenile Dermatomyositis

Developed by Josh Koegler and Dr. Dax Rumsey for PedsCases.com.
March 27, 2018

Introduction:

Hi, welcome to Pedscases. My name is Josh Koegler and I am a third year medical student at the University of Alberta working with Dr. Dax Rumsey, a Paediatric Rheumatologist at the Stollery Children's Hospital in Edmonton, Alberta. The topic for today is an approach to juvenile dermatomyositis, or JDM. Dr. Rumsey and I have worked together to bring you this podcast and we hope you will find it useful.

Objectives

By the end of this podcast, you should be able to:

1. Generate a differential diagnosis for a child presenting with weakness
2. Describe key points on history for a child presenting with JDM
3. Demonstrate a focused physical exam on a child presenting with JDM
4. Identify key investigations for JDM
5. Gain a basic understanding for the treatments and prognosis for JDM

Background Info

Juvenile Dermatomyositis (JDM) is a disease that involves myositis – inflammation of muscles – that classically presents as proximal weakness. Besides myositis, there is also a key dermatologic component of characteristic rashes, hence the name dermato – myositis. In addition to these key findings, there are other findings and complications associated with JDM which will be discussed. JDM affects about 4 in 1 million children per year, and is more common in females. The cause of JDM is unknown, but it is believed to be an autoimmune condition, likely caused by environmental triggers in genetically susceptible individuals, where cells of one's own immune system invade muscles and skin causing inflammation.

Now that we have some background information on JDM, let's get to our case!

Case

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You are a medical student on your general paediatric rotation. You've been asked to see a 6-year-old girl named Mya in clinic today. You glance at the nurse's note and see that Mya is presenting with a rash and progressive weakness with difficulty in her day to day tasks.

DDx

Although weakness is one of the key features of JDM, many other conditions can cause weakness and JDM is rare, so it is important to start out with a wide differential for a child presenting with weakness. Additionally, in JDM the weakness (and rash) may be subtle at first, which can result in a delay in diagnosis. So, it is important to reliably identify and quantitate weakness. Before we go over possible causes of weakness, take a moment to consider your own differential diagnosis for a child presenting with weakness. What conditions would you be considering?

Other causes of weakness include viral-associated causes (such as influenza, coxsackievirus B, and many others), Guillain-Barre syndrome (which presents as rapid onset ascending weakness, most commonly triggered by infection), myasthenia gravis (which is an autoimmune condition known for fatigable weakness), muscular dystrophies (such as Duchenne and Becker), endocrinopathies (such as hyperthyroidism, hypothyroidism, Cushings and Addisons), mitochondrial myopathies, metabolic disorders such as glycogen and lipid storage diseases, and polymyositis (another autoimmune disease which, unlike dermatomyositis, is not associated with skin findings and is rare in children).

Now that we have a basic differential diagnosis for a child presenting with weakness, let's go through some crucial questions we don't want to miss on history! Take a moment to consider what questions you would like to ask Mya.

History

Let's go through some important questions to add to your basic history:
A. History of Presenting Illness

1. Weakness
It's important to characterize the weakness. Ask about the onset, and duration, as well as which muscles are most affected which may be more easily elucidated by asking about which activities have become most challenging.
In JDM, the weakness is typically symmetrical and involving the proximal muscles, including the neck flexors, shoulder girdle muscles, and hip flexors. So, ask about activities which could be impeded by weakness in these muscles, such as climbing stairs, combing hair, or even getting out of bed.
It's important to note that JDM can cause weakness in muscles used for speaking, swallowing, and breathing which can be very dangerous and is important to assess. Remember the 3 Ds - dysphonia, dysphagia, and dyspnea. If any of these are present, then you should be more concerned. To help rule out other causes of weakness, ask about the

child's family history to help identify hereditary causes of weakness as in muscular dystrophy, ask about fatigable weakness – which can be seen in myasthenia gravis, and ask about current or recent infections which can also lead to weakness.

2. Rashes
Ask about any new rashes, especially around the eyes, and on the finger joints, elbows, knees, or ankles. As well, inquire about any skin ulcerations and skin changes such as hardening or calcinosis (i.e. calcium deposits in the skin).

3. Associated symptoms
It's important to identify associated symptoms such as arthritis, muscle tenderness, and fatigue.
Ask about gastrointestinal symptoms including abdominal pain and GI bleeding, as well as cardiovascular symptoms such as chest pain.

4. Constitutional symptoms
Ask about fevers and weight loss, which can both occur in JDM. Although Dermatomyositis can also be associated with malignancy that association is mainly found in adults with dermatomyositis and is extremely rare in juvenile dermatomyositis.

To summarize, JDM typically presents with rash, weakness or both, and can have additional findings such as fevers, arthritis, muscle tenderness, and fatigue. Warning symptoms to look out for include voice change, trouble swallowing, and difficulty breathing. Rarely, gastrointestinal tract or cardiac involvement may be present.

Case

Let's get back to our case. You take a thorough history and discover that Mya's rash involves her hands and face. Her parents initially thought it was an allergic reaction and were given a lotion at another clinic that has not helped. As for her weakness, it's hard for Mya and her parents to describe when it started. By asking activity-related questions, you find that over the past month Mya seems to have had more difficulty than usual with activities that involve her proximal muscles (e.g. getting up from sitting, doing her hair, and going up and down stairs). Mya hasn't had any fevers or weight loss, and doesn't describe any issues with breathing, speaking, or swallowing (i.e. the 3 Ds).

Now that we are satisfied with the history, let's move on to the physical exam:
But first let's take a moment to consider key findings that may inform your diagnosis.

Physical Exam

It's important to record the patient's vital signs, as well as their height and weight, and to plot these, noting any changes in growth over time.
In addition to your basic physical exam, we also want to assess muscle weakness. Important signs that may be observed include signs of weakness of the neck flexors (in the worst case, inability to lift the head off the exam table), Trendelenburg sign showing hip

ABductor weakness (a positive sign is if, when the child is asked to stand on one leg, his/her contralateral hemi-pelvis drops), and Gower's sign showing proximal muscle weakness (where in order to stand up the child supports their weight by bracing and moving their hands up their thighs). Additionally, the patient may not be able to sit up from a supine position, and instead may roll onto their side.

As discussed, the weakness can include muscles used for speaking, swallowing, and breathing leading to dysphonia, dysphagia, and dyspnea. You should check the strength of all the major muscle groups by having the patient resist counter-pressure that you apply. In the most extreme cases of weakness, the patient may not even be able to resist gravity. Complete a joint exam and a thorough skin exam. Assess for any skin breakdown and skin changes as JDM can cause ulcerations, as well as calcinosis. The characteristic rashes of dermatomyositis are Gottron's papules and heliotrope rash. Gottron's papules are papulosquamous lesions typically found over the knuckles. Similar lesions can also be observed on the extensor surfaces of several other joints, including the elbows and knees, as well as the medial malleoli of the ankles. The heliotrope rash is a reddish-purple rash on the upper eyelids, which is also often associated with upper lid edema.
Although those were wonderful descriptions, now would be a great time to pause the podcast and look up images of Gottron's papules as well as the heliotrope rash.
Additionally, a malar rash on the face (similar to in lupus) can be present. Other rashes include the shawl sign, which is an erythematous rash around the upper back, and the V-sign, a rash on the upper chest.

Look for capillary changes at the nail folds. Capillary changes are a common finding in JDM, and include changes such as vessel dilatation, tortuosity (twisting), and dropout (which means fewer capillaries than normal are observed).
When performing the chest exam, look for signs of cardiac involvement such as the pericardial friction rub of pericarditis, which, thankfully, is rare in JDM.
During the abdominal exam, look for abdominal distension, guarding, and rigidity and be aware that although associated vasculitis of the GI tract is rare, it can cause severe manifestations, including bleeding, perforations, and infarctions.
During your pulmonary exam, be aware that muscle weakness and/or pulmonary fibrosis may cause decreased chest expansion.

Case

Back to our case: You perform a physical exam and find significant weakness of Mya's hip, shoulder and neck muscles. Her distal muscles seem to be spared, and her reflexes, and sensations are intact. Cardiac, respiratory, abdominal and cranial nerve exams are unremarkable. On closer inspection of Mya's rash, you do note the characteristic heliotrope rash around both of her eyes, and Gottron's papules over her MCPs, PIPs, and DIPs of both hands. You don't appreciate any other rashes or capillary changes.

Now that you have an approach to the history and physical, let's take a moment to identify the diagnostic criteria for juvenile dermatomyositis and the investigations we can use to confirm our diagnosis.

Diagnostic Criteria and Investigations

The diagnostic criteria most often used are those by Bohan and Peter. However, these criteria were made in 1975, before most of us were even a twinkle in our parents' eyes! Currently, new criteria for JDM are being developed and should be available soon. Nevertheless, the existing criteria include the classic rash (heliotrope rash or Gottron's papules), proximal and symmetrical muscle weakness, elevation of muscle enzymes (including creatine kinase, lactase dehydrogenase, aldolase, and aspartate aminotransferase), electromyographic (EMG) changes, and a muscle biopsy showing characteristic changes. A probable diagnosis includes the classic rash plus two other criteria. A definite diagnosis includes the classic rash plus three other criteria.

The above criteria highlight important investigations, including serum levels of muscle enzymes, muscle biopsy, and electromyography. However, nowadays EMG and biopsy are not often used in clear cases of JDM. Although not included in the 1970s criteria, MRI is now a useful tool in documenting the presence and extent of muscle inflammation, while EMG and biopsy are usually reserved for atypical cases or those that do not respond well to typical therapy. Additionally, a myositis autoantibody panel can be sent, which can help inform prognosis.

Pulmonary Function Tests can help identify a restrictive defect with pulmonary involvement, and swallowing assessment including contrast studies can be used to assess dysphagia and risk of aspiration. EKG +/- echo is often done at diagnosis to look for cardiac involvement.
Plain x-rays can also be useful in identifying the extent of calcinosis.

However, as a general paediatrician or family doctor, the key step in a JDM workup is a prompt consultation with a paediatric rheumatologist as soon as JDM is suspected (e.g. child with muscle weakness and rash).

Case

You and your preceptor discuss the case and decide JDM is the most likely cause of Mya's presentation. You both discuss with Mya and her parents the initial plan to order muscle enzymes and promptly refer Mya to a paediatric rheumatologist. Mya's parents are quite anxious, but relieved that there might finally be an answer to what Mya has been experiencing. They have never heard of anyone having JDM and are very curious about what treatment options.

Treatments

The two main medicines used to treat JDM in 2017 include corticosteroids and methotrexate. Prior to corticosteroids, 1/3 of children with JDM died from their disease. Methotrexate is often used as a steroid-sparing agent. This is done because corticosteroids are wonderfully powerful drugs, but they often have unwanted side effects such as increased appetite, mood changes, osteoporosis, hypertension, and many more. Hence,

the addition of a steroid-sparing agent such as methotrexate allows a lower dose of steroid, while still maintaining a desirable response. To use a simple analogy, we use steroids to put out the fire of inflammation and methotrexate to keep it out. If the presentation is severe and/or you suspect GI involvement and, thus, decreased absorption of oral medications, you can give the steroids intravenously at first. Vitamin D and Calcium are also given to counteract negative effects of steroids on bone health.
Additionally, Intravenous Immunoglobulin (IVIG) can be given in severe unresponsive disease (particularly for resistant rashes).

Alternative treatments include: other disease modifying anti-rheumatic drugs (DMARDs) besides methotrexate, biologics, and cytotoxic agents. These treatments are for those children who either do not tolerate standard treatment, do not respond to it, or have another contra-indication(s) to its use.

Other key elements of treatment include physiotherapy, occupational therapy, dietician involvement, and social work support. Overall, a coordinated healthcare team is critical in optimizing the health and function for children with JDM.

Be aware that children with JDM may need to be admitted to hospital if they present with severe weakness (remember the 3 Ds - dysphagia, dysphonia, dyspnea), aspiration pneumonia, severe major organ system involvement (e.g. GI ulcerations or perforation), or secondary infection.

Case

The paediatric rheumatologist sees Mya, completes the work-up and confirms the diagnosis of JDM. He starts treatment to help get Mya's condition under control. He is impressed with your team's work-up, prompt recognition of the condition, and appropriate referral. Several months later, you are doing a rotation in paediatric rheumatology and you see Mya back in clinic. Mya's parents are happy to tell you things have settled down, but they seem concerned about her future. You've read up on JDM since your paediatric rotation and are ready to answer some of their questions.

Complications and Prognosis

As mentioned above, specific antibodies – known as myositis specific antibodies can aid in prognosis. Of children diagnosed with JDM, about 1/3 will have will have a monocyclic course (in that they will recover from their disease), about 1/3 will have a polycyclic course (in that they will have disease-free periods followed by relapses), and 1/3 will have a chronic continuous course. Mortality has remained low (95% long- term survival) since the introduction of corticosteroids for treatment of JDM. The main long-term complications of JDM are calcinosis, lipodystrophy, contractures and other metabolic sequelae (such as diabetes, insulin resistance, and hyperlipidemia). Thus, patients need to be followed long-term. Overall, with today's treatments most patients do generally well if treated early and appropriately.

Case Conclusion

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We've been able to see Mya through the beginning of her journey from presentation to counselling on prognosis and hopefully have learned some things about juvenile dermatomyositis along the way. Let's end our discussion with some key take home points.

Take-Home Points

1. Remember to consider a wide differential diagnosis for a child presenting with weakness.
2. If JDM is suspected during your history, don't forget to assess weakness and function with activity-related questions, and don't forget to ask about and assess the 3 Ds – dysphagia, dysphonia, and dyspnea.
3. Your JDM-focused physical exam should include a reliable weakness assessment, thorough skin and joint exam, and a full systems exam to identify any cardiac, lung, or abdominal complications.
4. Muscle enzymes are part of the general work-up for a child with suspected JDM but remember a prompt paediatric rheumatologist referral is key.
5. Lastly, remember that JDM treatments are mainly focused around corticosteroids and DMARDs with a multidisciplinary healthcare team to meet the individual needs of patients. Some patients will have a monocyclic course, some will have relapses and periods of remission, and some will have a chronic, difficult to treat course, but most patients do generally well if treated early and appropriately.

Well, we've reached the end of our podcast for today's PedsCase on Juvenile Dermatomyositis. We hope you have a better understanding of this condition and an interest to learn about other topics provided on PedsCases.com. Thanks for listening!

References

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