Social Media in the Rare Disease World

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Fine Print

- I have no conflicts of interest to declare
- Nothing presented should be considered legal advice
- Nothing presented necessarily represents the views of the College of American Pathologists or its Board of Governors

Pulmonary and Ophthalmic Involvement With Erdheim-Chester Disease

A Case Report and Review of the Literature

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• **Erdheim-Chester disease is a rare nonfamilial histiocytic disorder of unknown etiology with characteristic long bone findings. The 3-year survival rate for patients with Erdheim-Chester disease is 50%. Approximately 50% of patients have disease involvement in other tissues, including skin, retro-orbital and periorbital tissues, pituitary-hypothalamic axis, heart, kidney, retroperitoneum, breast, skeletal muscle, and sinonasal mucosa; about 20% of patients have lung involvement. Prognosis generally depends on the extent of the extraosseous disease. For patients with lung involvement, gender distribution is equal, but men typically present at an older age than do women. Approximately 80% of patients present with dyspnea, and most patients have diffuse interstitial infiltrates and pleural and/or interlobar septal thickening on chest radiology. Characteristic lung histopathology includes the accumulation of histiocytes with variable amounts of fibrosis and a variable lymphoplasmacytic infiltrate in a lymphangitic distribution. Immunostains are diagnostically useful, showing immunopositivity for CD68 and factor XIIIa and immunonegativity for CD1a. Birbeck granules are uniformly absent ultrastructurally.**

(*Arch Pathol Lab Med.* 2004;128:1428–1431)

Erdheim-Chester disease is a rare nonfamilial histiocytic disorder first identified by William Chester in 1930 that primarily affects middle-aged and older adults and predominantly involves the long bones of the extremities.[1–3] Bone pain is the most common presenting symptom, and characteristic radiographic changes in the long bones—bilateral cortical sclerosis predominantly involving the metaphyses and diaphyses—is considered virtually pathognomonic.[2] The etiology of Erdheim-Chester disease is a monoclonal proliferative disorder or whether it is a polyclonal reactive disease.[4,5] While Erdheim-Chester disease has been known to occur in patients who also have Langerhans cell histiocytosis, the 2 diseases are generally considered to be separate entities.[4] Neither has Erdheim-Chester disease been found to be a lipid storage disease.[5] Approximately 50% of patients have disease involvement in other tissues, including skin, retro-orbital and periorbital tissues, pituitary-hypothalamic axis, heart, kidney, retroperitoneum, breast, skeletal muscle, sinonasal mucosa, and lung.[4] Prognosis generally depends on the extent of the extraosseous disease, with 59% of patients reportedly dying of their disease and 36% dying of the disease at 6 months.[2,6]

One hundred seventy-six cases of Erdheim-Chester disease have been reported in the literature. One hundred sixty-four cases had been reported as of August 2002,[4] and our review of the English-language literature shows 12 additional case reports published between August 2002 and June 2003.[7–18] Forty-one (23%) of the 176 cases showed pulmonary involvement. Detailed descriptions of the pulmonary histopathology were present for 23 of the 41 cases with pulmonary involvement (Table). This case report represents the 24th reported case of Erdheim-Chester disease with pulmonary involvement that presents a detailed description of the pulmonary histopathology.

REPORT OF A CASE

A 60-year-old man with no history of smoking and a past medical history of hypertension presented with painless loss of vision in one eye, dyspnea, iron deficiency anemia, decreased renal function, and sclerotic and lytic lesions on long bone x-ray. Prominent slightly elevated yellow plaque lesions were present on all 4 eyelids (Figure 1). Well-circumscribed lesions surrounding the

able S100 immunostaining pattern may be attributable to reactive histiocytes, which generally are S100 immunopositive, infiltrating the fibrohistiocytic areas in reaction to the Erdheim-Chester disease. Further examination of these S100-immunopositive histiocytes may be helpful in determining whether they are reactive histiocytes, histiocytes of Erdheim-Chester disease, or histiocytes of another origin. Ultrastructural studies were performed in 6 cases; none contained Birbeck granules, typically associated with Langerhans cell histiocytosis.

In summary, Erdheim-Chester disease is a rare nonfamilial histiocytic disorder of unknown etiology with characteristic long bone findings. Overall, the 3-year survival rate in Erdheim-Chester patients is 50%. About 1 in 5 patients have lung involvement. Our study found that for these patients, the 3-year survival rate was 66%. For patients with lung involvement, gender distribution is equal, but men typically present at an older age than women. Four of five patients present with dyspnea, and most patients have diffuse interstitial infiltrates and pleural and/or interlobar septal thickening on chest radiology. Characteristic lung histopathology includes the accumulation of histiocytes with variable amounts of fibrosis and a variable lymphoplasmacytic infiltrate in a lymphangitic distribution. Immunostains are diagnostically useful, showing immunopositivity for CD68 and factor XIIIa and immunonegativity for CD1a. Birbeck granules are uniformly absent ultrastructurally.

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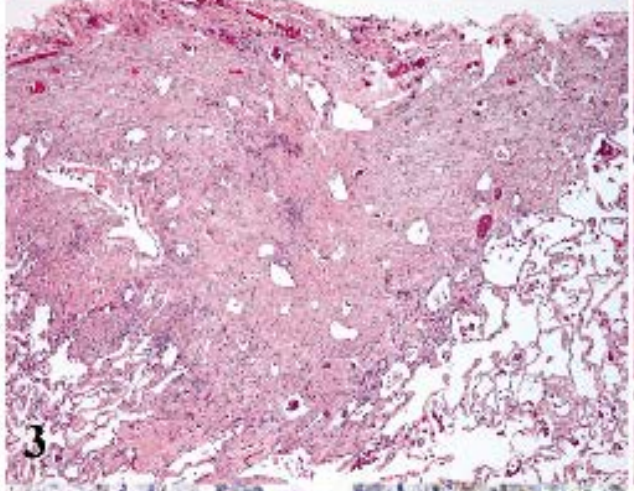

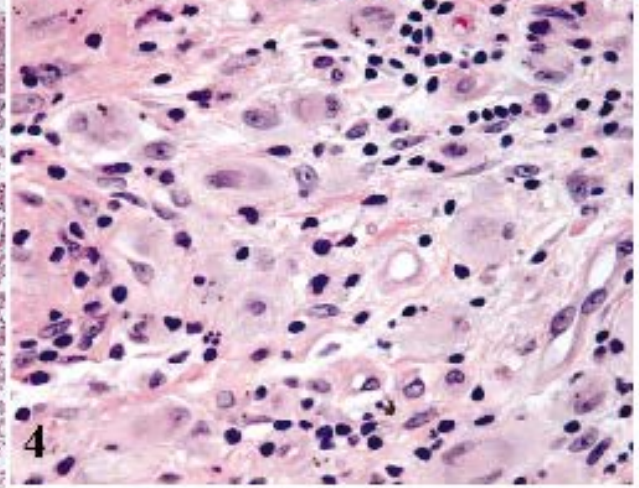

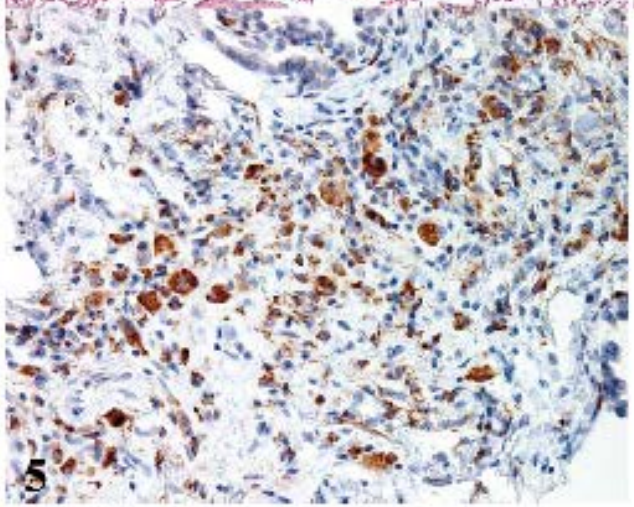

Figure 1. *Yellow plaques present on all eyelids.*

Figure 2. *Orbital mass biopsy showing adipose tissue with fibrosis, a mixed inflammatory infiltrate, and interspersed histiocytes (hematoxylin-eosin, ×100).*

Figure 3. *Subpleural mature fibrosis with a variable lymphoplasmacytic infiltrate and histiocytes (hematoxylin-eosin, ×40).*

Figure 4. *Higher power, showing histiocytes in lung biopsy (hematoxylin-eosin, ×400).*

Figure 5. *CD68 immunostaining of histiocytes in lung biopsy (hematoxylin-eosin, ×200).*

- Facebook Page:
- https://www.facebook.com/ErdheimChesterDisease
- Facebook Group
- https://www.facebook.com/groups/784637751599933/

Twitter

Erdheim-Chester
@ECDGA FOLLOWS YOU

- TWEETS 233
- FOLLOWING 135
- FOLLOWERS 79
- FAVORITES 19

Why Social Media?

- The Pathologist
  March 3rd, 2015 Issue #0215
  Author: Fedra Pavlou

- “Social media could help. For me, Queen Rania of Jordan – a somewhat unexpected social media guru – sums up both the equalizing nature and the potential power of modern communication tools: ‘Social media are a catalyst for the advancement of everyone’s rights. It’s where we’re reminded that we’re all human and all equal. It’s where people can find and fight for a cause, global or local, popular or specialized, even when there are hundreds of miles between them.’”
  - https://thepathologist.com/issues/say-what/tweet-up/

Janet Freeman-Daily @JFreemanDaily

- EPATIENTS ON THE FRONT LINES: PRECISION MEDICINE, THE FDA, AND ME
- On February 19, 2015, I was an invited patient advocate speaker at the 11th Annual Moores Cancer Center Industry/Academia Translational Oncology Symposium. My topic, “EPatients on the Front Lines: Precision Medicine, the FDA, and Me,” explained how cancer research could move faster and be more successful if researchers, pharmaceutical companies, and the biotech industry would collaborate with patients early in the trial design process.
  - http://grayconnections.net/2015/02/20/epatients-on-the-front-lines-precision-medicine-the-fda-and-me/

@JFreemanDaily

#lcsm

- “I was diagnosed with Stage 3a non-small cell lung cancer in May 2011. I never smoked anything – except a salmon.”
- “However, I wasn’t just a recipient of care. The information I learned in the Inspire online lung cancer community enabled me to become an interactive participant. From other epatients, I learned to ask for my data, including radiology and pathology reports. I also learned more extensive molecular testing was available at other facilities, and arranged to have my slides sent to the University of Colorado Hospital for a 10-oncogene panel. Unfortunately, all tests were negative.”

How is an Erdheim-Chester Disease Patient Like a Pathologist?

- HINT:
- “One hundred seventy-six cases of Erdheim-Chester disease have been reported in the literature. One hundred sixty-four cases had been reported as of August 2002,4 and our review of the English-language literature shows 12 additional case reports published between August 2002 and June 2003.7–18 Forty-one (23%) of the 176 cases showed pulmonary involvement.”

- Timothy Craig Allen, Patricia Chevez-Barrios, Debra J. Shetlar, and Philip T. Cagle (*2004*) Pulmonary and Ophthalmic Involvement With Erdheim-Chester Disease: A Case Report and Review of the Literature. Archives of Pathology & Laboratory Medicine: December 2004, Vol. 128, No. 12, pp. 1428-1431.

Allen TC. Social Media: Pathologists' Force Multiplier. *Arch Pathol Lab Med*. 2014 Aug;138(8):1000-1.

- "Pathologists…make up only about 0.006% of the population of the United States."
- "Pathologists cannot sway legislators solely by appealing as a numerically powerful constituency; nor can we sway other policymakers, including administrative leaders and quasi-legal agencies, by force of sheer numbers…Lots of people do not even know pathologists are physicians."
- "The goal of sharing our message with patients, colleagues, the public at large, policymakers, and even an international audience cannot be met solely from face-to-face encounters."
- "To educate the public about pathology, and to meaningfully engage in and affect health care policy, pathologists need something more—a tool to help us overcome the seemingly insurmountable limitation of our numbers. Pathologists need something not typically considered in pathology. Pathologists need a force multiplier."

6 Types of Social Media

- **Social networks** (Facebook, LinkedIn) *connect to others with similar interests*
- **Bookmarking sites** (Delicious, StumbleUpon) *save, manage sites, etc.*
- **Social news** (Digg, Reddit) *links, news posted; people vote as core social aspect*
- **Media sharing** (YouTube, Flickr) *video, pictures, with ability to comment*
- **Microblogging** (Twitter) *short items or updates to subscribers*
- **Blog Comments and Forums** (lots of them) *online forum members have conversations via message posting; bloggers post on topics, people comment*

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Twitter

- The Beginner's Guide to Twitter
- http://mashable.com/2012/06/05/twitter-for-beginners/#:eyJzIjoidCIsImkiOiJfc3NiMzhoeHE1bTcxOXdqbCJ9
- CAP: A crash course of twitter
- ttps://www.youtube.com/watch?v=w5pz0kAMVi4
- CAP: How to twitter youtube
- https://www.youtube.com/watch?v=-vefcZL6w80sdfg
- CAP: 10 tips to a good tweet
- https://www.youtube.com/watch?v=V6zlUaYoaE8

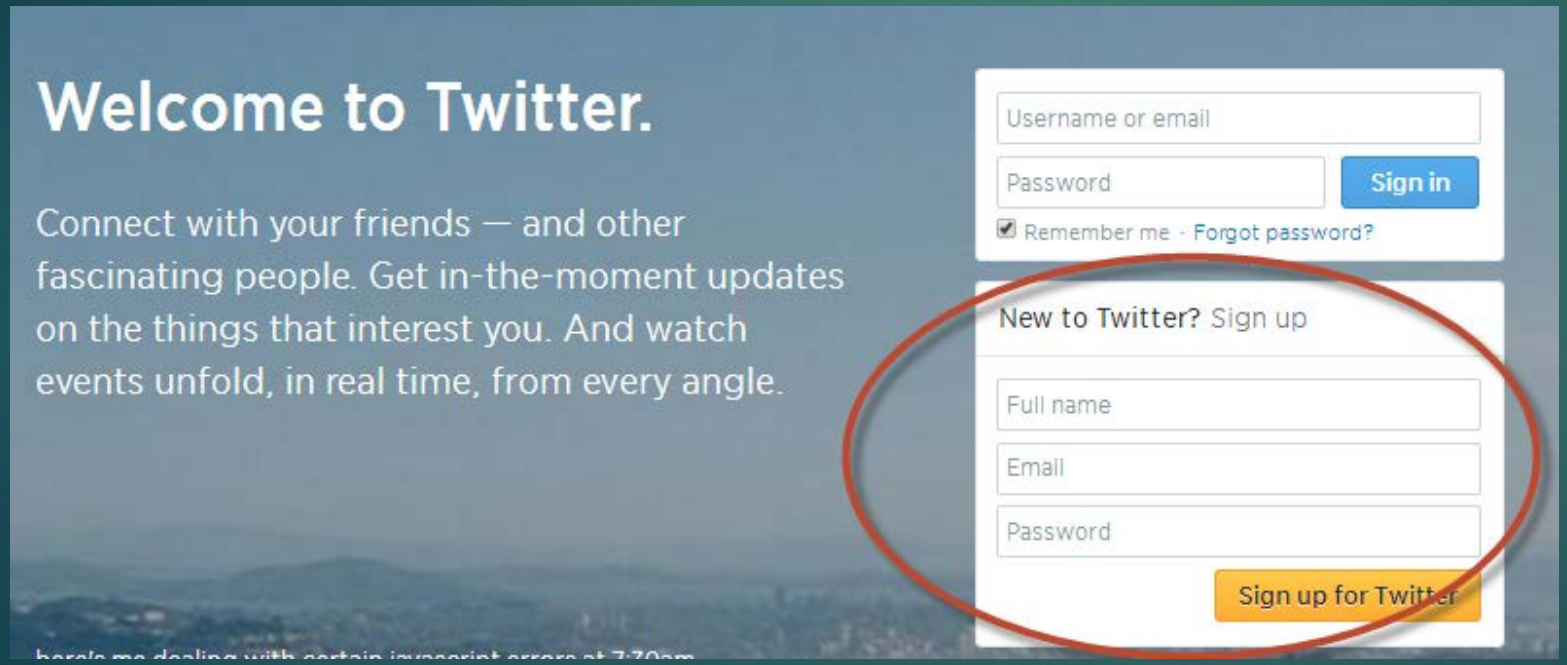
- https://www1.gotomeeting.com/register/423726697

Twitter

- Launched: April 2007
- Online social networking and microblogging service that enables users to send and read short 140-character text messages, called "tweets"
- Users access Twitter through the website interface, SMS, or mobile device app
- Currently 255 Million active users, 57 million in the USA
- 500 million Tweets per day
- 78% of Twitter active users are on mobile
- 77% of accounts are outside the USA

How to establish a Twitter account and create a profile

Go to: www.twitter.com

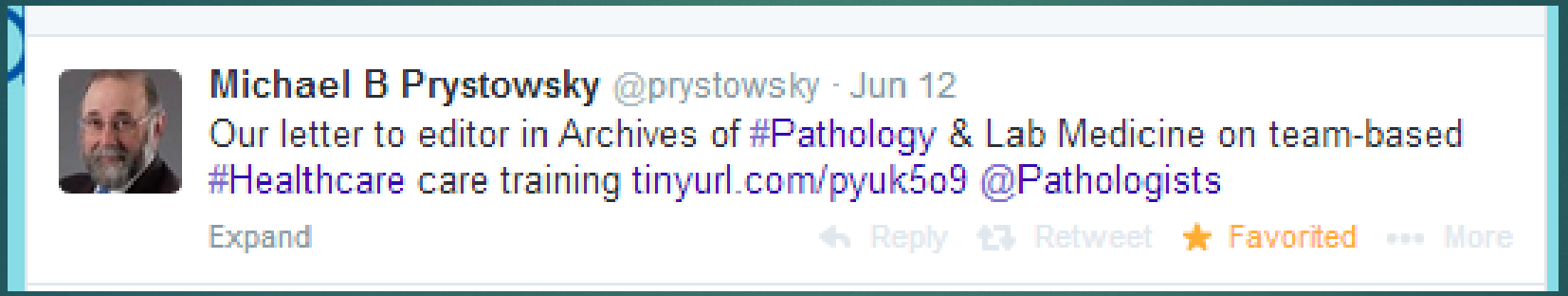


Tweeting and # (hashtags)

- Tweets are short messages,140 characters or less
- Use comma or “&” instead of “and”
- Use dash, abbreviations; remove spaces
- Hashtag (the # symbol) marks keywords or topics in a Tweet; allows one to find and follow a category or subject
- Putting “#” in front of a word or phrase (no spaces) in a Tweet allows them to be easily searched
- Clicking on a hashtagged word in any message shows you all other Tweets marked with that word or phrase
- Hashtags can occur anywhere in the Tweet – at the beginning, middle, or end; and are often used to denote Trending Topics

Replies, favorites, and mentions

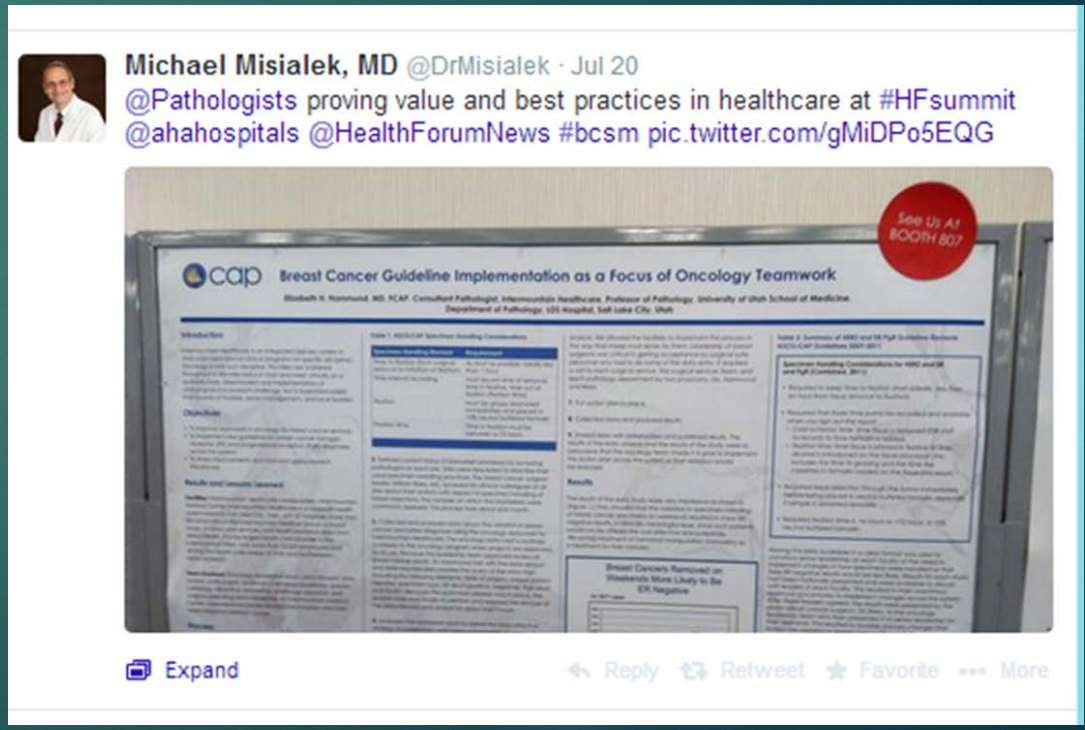
- A “mention” is a tweet that contains "@username"
- Replies using “@username are also mentions



Engage!

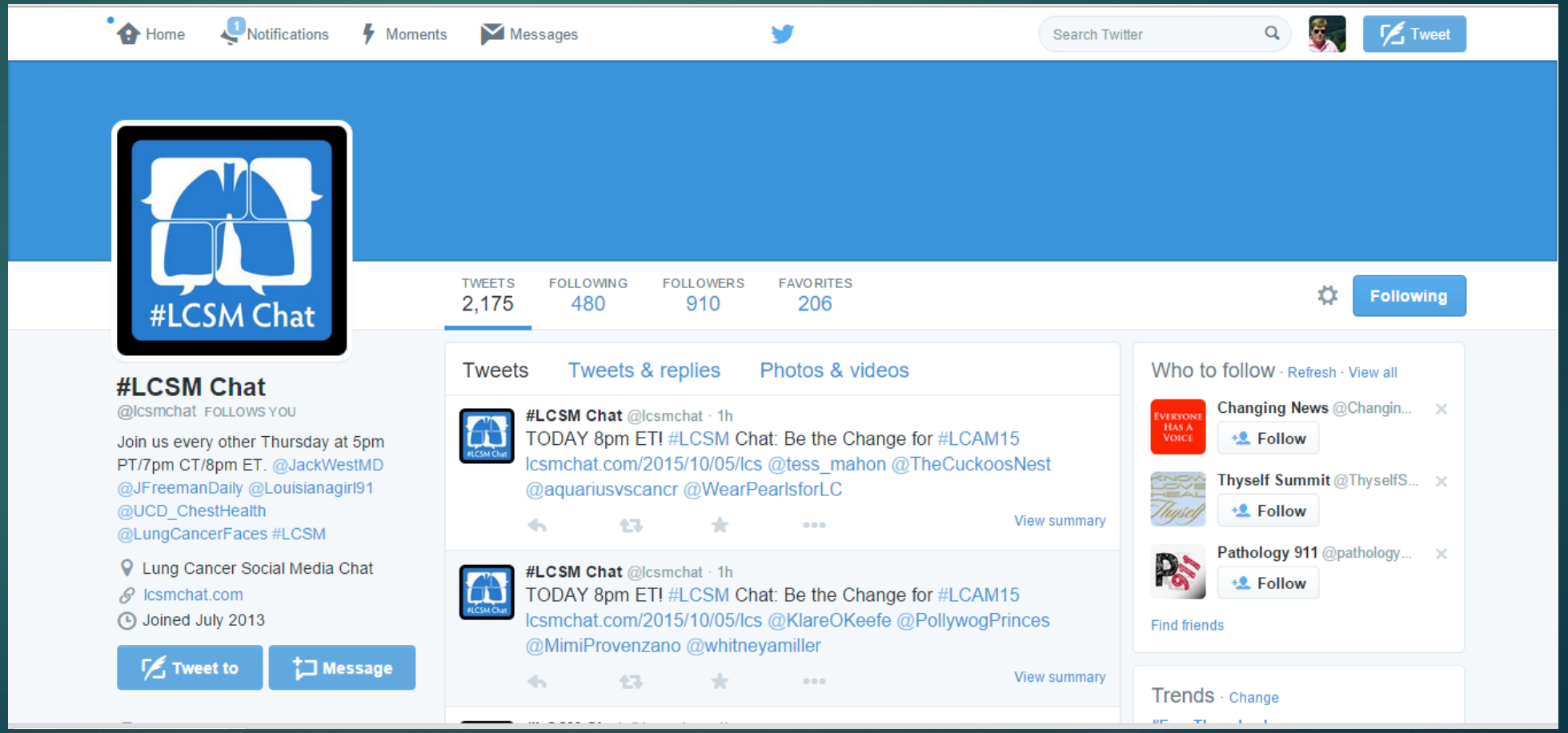
- Ask questions

- Tweet from (and about) meetings



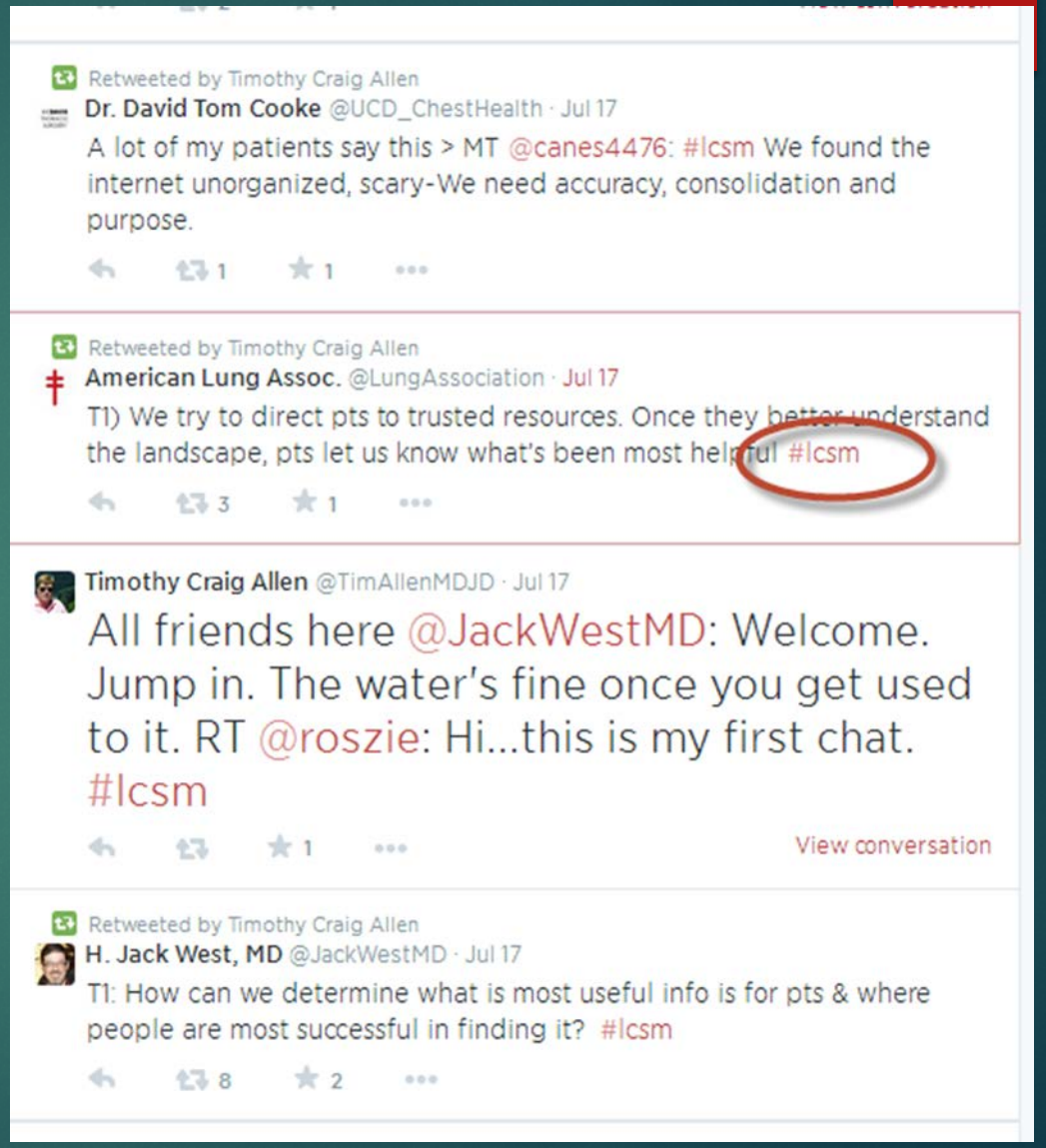
Engage!

Twitter Chats



Engage!

- Twitter chats: direct involvment
- Blogs—free platforms online; leverage blog posts to Twitter, Facebook, Google+ http://timallenmdjd.blogspot.com
- All of these Influence other patients, other families, physicians, policymakers



Optimize

- Use links
- Retweet
- Cite journal articles
- Ask questions
- Go mobile
- Leverage yourself; add twitter handle to emails, facebook posts

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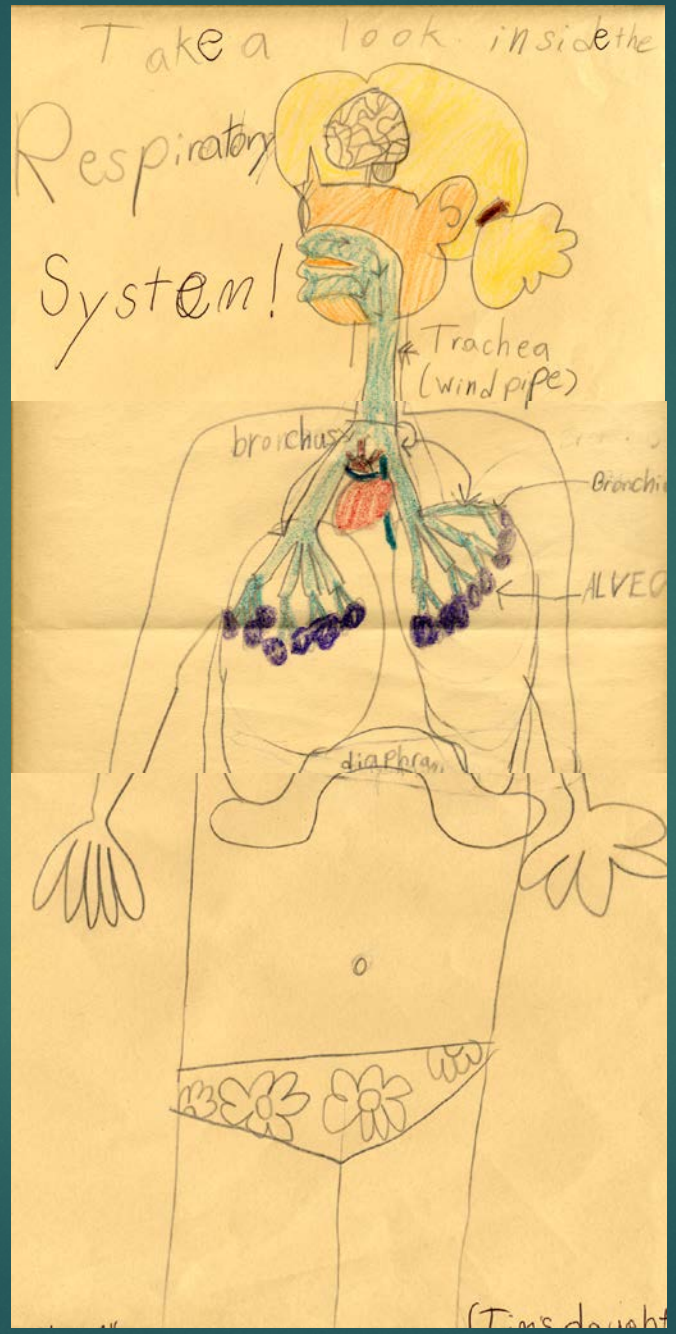
Take a look inside the
Respiratory
System!
Trachea
(wind pipe)
bronchus
Bronchi
ALVEO
diaphragm