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Cystic Fibrosis in Adults: Why you should keep it on the differential

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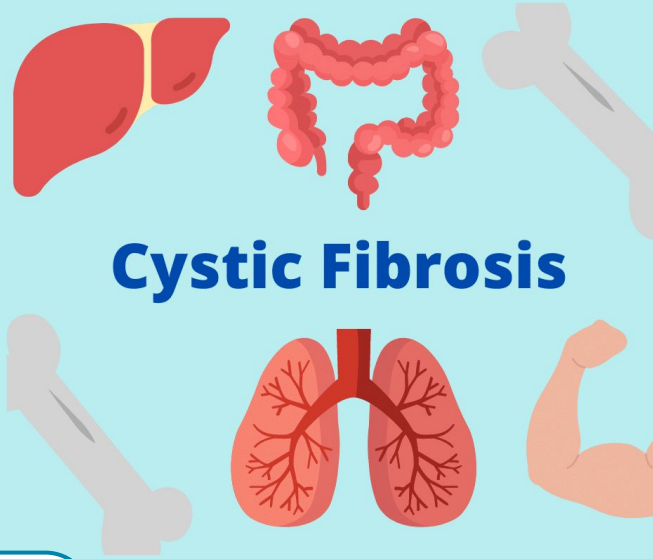
Background

- Cystic Fibrosis (CF) occurs from mutations in the Cystic Fibrosis transmembrane conductance regulator (CFTR) gene resulting in thick secretions that affect multiple organs.[1]

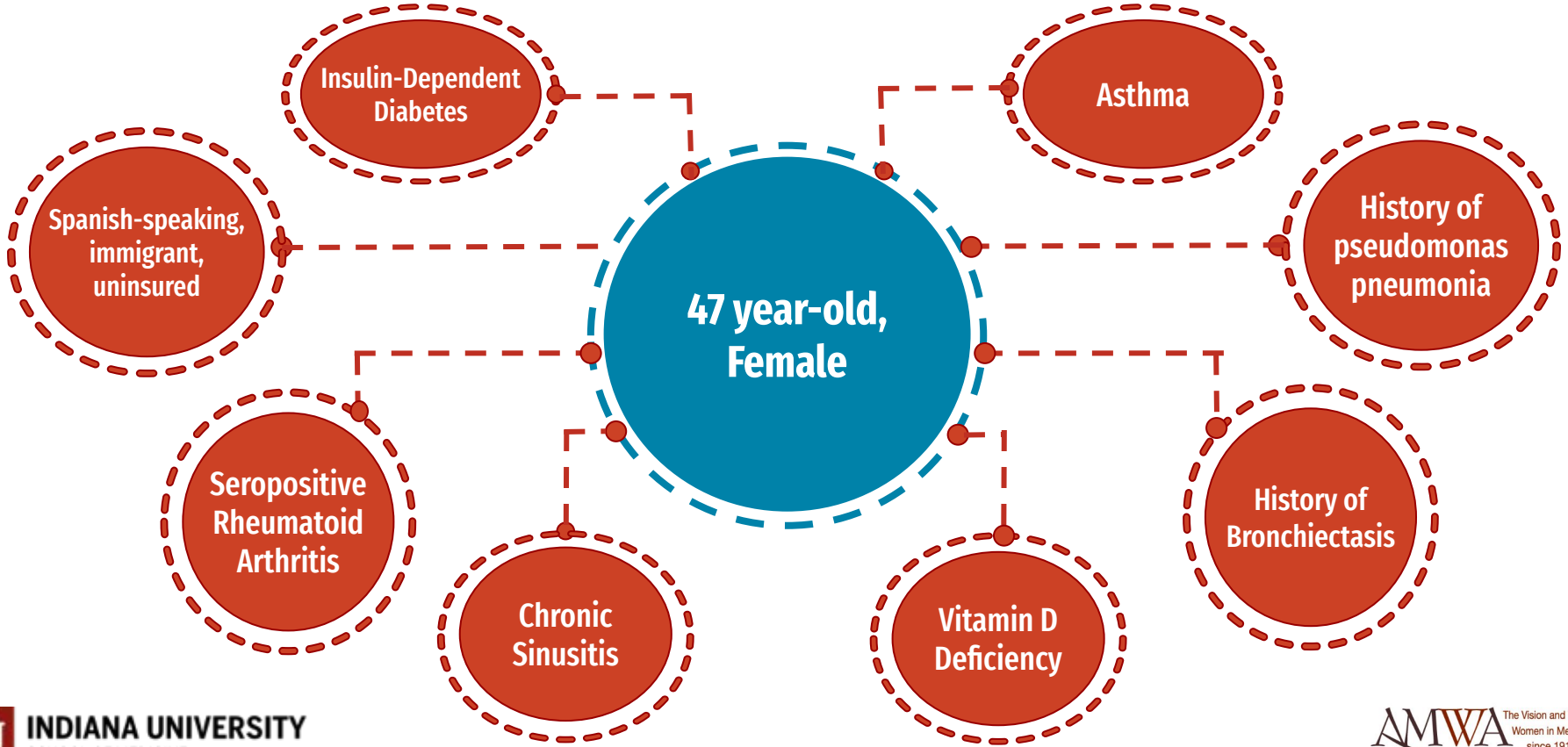
- Diagnosis of CF in adulthood is often due to uncommon mutations that result in atypical presentations.[1,2,3]

- Diagnosis[1]:

Typical Features or History of CF in a Sibling or Positive NBS Test	+	Elevated Sweat Chloride Test or Two CFTR gene Mutations or NPD Testing Abnormality



Meet the Patient



Case Presentation

2013

November: Patient hospitalized for worsening cough, body aches, nausea, and vomiting

Bronchiectasis on CT

BAL revealed GNRs, likely pseudomonas given history of prior pseudomonas pneumonia earlier that year (Feb.)

- CFTR 32 gene panel

2015

September: Seen in pulmonology clinic for mild-persistent asthma

Stable, advised to follow-up in 6 months

2016

March: Presents to clinic with worsening cough

PFTs: FEV1=70%

+ Sweat Chloride test on two separate occasions, 77 and 81

September: Genetic testing revealed heterozygous for CF (p.Arg352Gln(a))[4]

Patient started on Creon and vitamin supplementation due to concern for pancreatic insufficiency

2022

Early 2022: Patient started on Trikafta

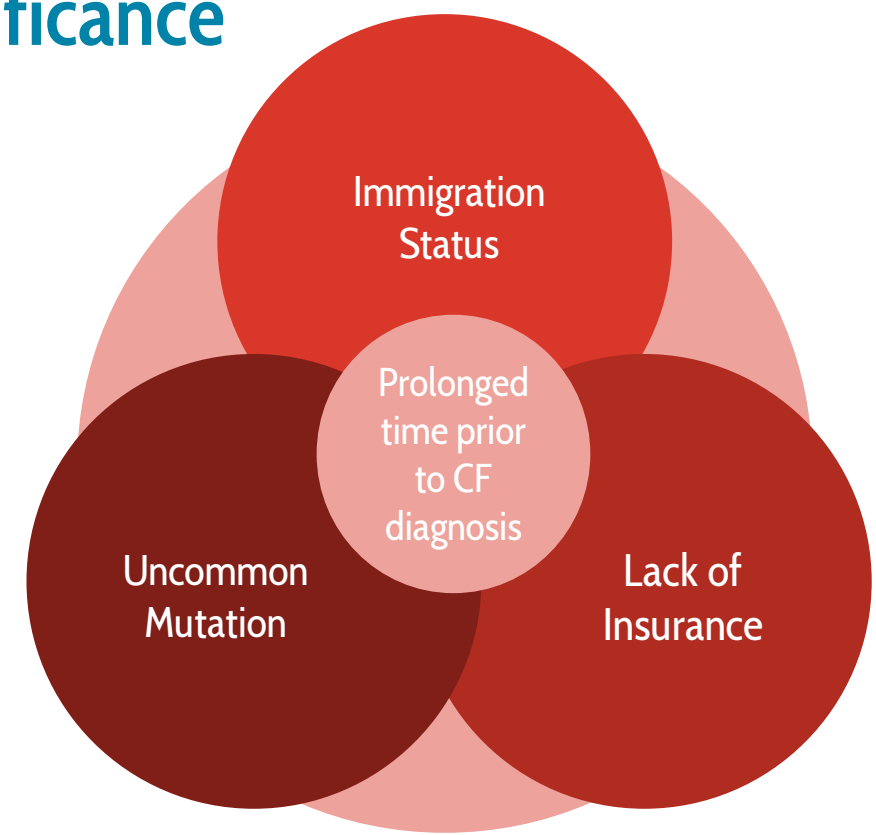
July: DEXA scan consistent with osteopenia

December: Hospitalized for CF exacerbation

2023

February: Patient started on Pulmozyme via Compassionate Use

Clinical Significance



Conclusion

Delay in diagnosis of CF is due to a combination of phenotypic variations and a variety of nonclinical factors.[2]

Life expectancy is significantly decreased in uninsured patients with known CF,[5] and likely further decreased when a diagnosis is delayed by additional barriers.

Due to a less severe and atypical presentation, patients with CF diagnosed in adulthood may present to different disciplines and receive several individual diagnosis prior to a CF diagnosis.[3]

Providers should have a high index of suspicion for CF in an adult who presents with signs consistent with CF.

References

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