The many faces of CF diagnosis, when to think of CF?

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As medical students there were certain diagnoses one had to think of independently of facts otherwise one would forget. Syphilis, SLE and Hodgekins were classical examples in adult medicine. In paediatrics CF is also one of them and it is distressing how late diagnoses of CF are made with glaringly obvious symptoms and physical signs.

Medicine is not that difficult and is an exercise in pattern recognition. If something falls out of the usual pattern it ought to alert one not to keep blundering on treating them but to stop and think. As an example, asthma in the great majority of cases is dead easy to manage. If it isn’t, there are 3 choices: a) It isn’t asthma, b) It is asthma but they are not taking treatment or following advice, and c) The least likely option: they truly have severe asthma.

The following is a list of alerts:

A) Conjugated jaundice in the neonate.
B) Not regaining birthweight within 2 weeks and certainly within 3 weeks.
C) Coughing in a baby aged <4 months – when was the last time you saw a healthy baby doing that?
D) More than 4 dirty nappies per day (Upper 95% confidence interval is 4).
E) Finding an unexpectedly low serum sodium or albumin when investigating for something else and/or deranged urine electrolytes (particularly sodium <10 mmol/L) even desmopressin resistant diabetes insipidus
F) Features of zinc deficiency including acrodermatitis enteropathica, peeling paint dermatitis and even Kwashiokor type dermatitis.
G) Peripheral oedema. Yes of course it could be nephrotic syndrome, protein losing enteropathy, or Turner’s Syndrome but it could also be CF.
H) Rectal prolapse, 50% will be CF.
I) Nasal Polyps in someone age < 8 years is almost certainly CF, > 12 years a lower chance.
J) Cough with sputum. Children do NOT cough up sputum under 8 years of age.
K) Finger clubbing. I never want to see again a letter sent only 10 years ago from a psychiatrist back to a paediatric gastroenterologist saying ‘thank you for referring this 12 year old child with diarrhoea, low weight and clubbing’.
L) Persistent left lower lobe collapse in a 10 year old ‘asthmatic’. What were they thinking?
M) Aspergillus/ABPA/Aspergilloma. Very rare in paediatric asthma but is less discriminating in adults.
N) Pancreatitis
O) Facial nerve palsies

CF is principally a **clinical** diagnosis. A diagnosis can be made:
- A) With or without 2 abnormal genes identified,
- B) With or without an abnormal sweat test
- C) With or without abnormal nasal potential difference measurements
  **but probably not all 3**

The corollary is also true - that a child does not appear to have CF but:
- A) May have 2 abnormal genes identified
- B) May have an abnormal sweat test.
- C) May have abnormal nasal potential differences.
  **but probably not all 3**


