Fitting the patient to the disease:
Repeating Procrustes' mistake in a time of HIV infection.

Griessel DJ, MBChB MMed (Ped)
Schoeman CJ, MBChB MMed (Ped) DCH DCM
Henderson BD, MBChB MMed (Ped) DCH
Department of Paediatrics and Child Health
University of the Free State, Bloemfontein

Keywords: HIV, cystic fibrosis, infant, DNA

(SA Fam Pract 2005;47(5): 46)

Introduction
Procrustes was the mythical Greek innkeeper who either stretched or cut short his guests to fit a specific bed size. A classic mistake in medicine is to repeat this folly by adhering strictly to the "common things occur commonest" route. In a developing country with a high prevalence of HIV it is easy to ignore other possibilities, with consequences for the patient and family. We illustrate this point by presenting a case that was initially thought to fit the presentation of HIV disease but turned out to have another diagnosis, with a Mendelian inheritance pattern which made genetic counseling a worthwhile exercise.

Presenting history: A five-week old female infant presented with generalized oedema and skin rash of one week's duration. She was not thriving nutritionally although her milk intake was adequate.

Family history: The parents unrelated of Sotho origin are teachers. Two siblings of the patient died at age 4 months and 2 months respectively with a history of vomiting, diarrhoea and failure to thrive.

Past history: The patient was admitted to hospital at the third day after birth with abdominal distention and vomiting. The diagnosis of meconium ileus was made, was medically treated and discharged.

Special investigations:

FBC	Hb 8,6g/dl; WBC 13,2 x 10^9/l; Platelets 485 x 10^9/l
Electrolytes	Na 130, K 5, 0; Cl 108mmol/l
Urea	0,6mmol/l
Albumin	19g/l
Urinalysis	No abnormalities detected or organisms cultured
HIV antibodies	Negative
Sweat electrolytes	Insufficient sweat (to perform test)
chest X-ray	Normal

Physical examination: Growth parameters: Her weight and length were below the 3rd centile. She was pale with generalized oedema and desquamating hyper- and hypo-pigmented skin rash. The liver was palpable 3cm below the right costal margin

Definitive laboratory investigations:
1. Steatocrit : 70% (normal 0-25%)
2. DNA test for cystic fibrosis: Patient was homozygous for Cystic Fibrosis causing mutation 3120 +1GA

Discussion:
Cystic fibrosis (CF) is the most common life threatening recessive genetic disorder amongst populations of European descent. The disease is usually considered in patients with chronic lung disease or steatorrhoea. Until recently, very little was known about CF in populations of African origin, among whom it was believed to be extremely rare. It has now been shown that the carrier frequency of the abnormal CF transmembrane regulator gene in South African blacks is not uncommon and can be as high as 1 in 46.[2]

The incidence of CF in this population is predicted to be between 1 in 784 and 1 in 13,924 births. If this estimation is correct, many cases of CF in infants may be undiagnosed or misdiagnosed. There are several reasons for this, one of which is that CF can at times present with unusual clinical manifestations. The lesson of this short case study is to remind clinicians that CF does exist in the black population and that the presentation of the disease can be atypical.

An increased sweat sodium concentration is considered diagnostic of CF. This test is technically difficult in the very young and the presence of oedema is known to be associated with a false negative sweat test. This often leads to considerable delay in diagnosis. Dry skin and electrolyte disturbances also produce inaccurate results. A DNA test for CF is thus considered to be the investigation of choice in the young infant. The mutations found in the African population differ from those of European ancestry.[2] The laboratory should be informed to look for specific "African" mutations.

Conclusions:
A diagnosis of CF should be considered in black infants, who present with features of kwashiorkor (at an unusual age) or immune deficiency. Sweat electrolyte estimation is often unsuccessful in the very young or negative in the presence of oedema. DNA testing should be done to confirm the diagnosis of CF under such circumstances. *Everything that presents as a typical HIV infection is not necessarily HIV!*

References:
1. Pappworth, MH. A primer of Medi-cine. 3rd Edition, London: Butterworth 1971.
2. Padoa C, Golman A, Jenkins T, Ramsay M. Cystic fibrosis carrier frequencies in population of African origin. J. Med Genet 1999; 36: 41-44.