A rare case of erythrokeratodermia

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Case presentation

- 4 y.o. Hispanic male
- New patient being seen for “atopic dermatitis” that was resistant to many treatments over the past 3 years
- Mom reports rash initially started after a URI at age of 3 months
- Mom reports pregnancy was normal, with no family history of similar problems
Case continued

- Patient had one natal tooth, with rest coming in around 3 months of age
- Subsequently he has developed many caries, with weak abnormally shaped teeth, and an x-ray showed presence of permanent teeth
- Mom also reports nails fall off, he doesn't sweat a normal amount, and hair is fragile and falls out easily
- URIs exacerbate his rash, but no history of significant pulmonary infection, otitis media, cutaneous abscesses, warts, fungal infections
- Mom denies any other systematic problem
Biopsy

- Psoriasiform spongiotic dermatitis
  - Showed mild acanthosis along with mounds of parakeratosis and intervening orthokeratosis, with focally prominent hypergranulosis
Genetic testing

- Showed presence of desmoplakin mutation

Erythrokeratodermia cardiomyopathy syndrome

- Rare, with few cases reported
- Caused by dominant de novo mutation in desmoplakin
- Clinically have
  - Sparse hair, pruritic erythrokeratodermia
  - Hyperkeratotic, peeling, cracking palms and soles
  - Dental enamel defects, teeth abnormalities
  - Dystrophy of nails
  - Cardiac defects that develop at a young age
- Reported use of ustekinumab with great improvement
Our patient

- After about 1 year of treatment, patient still doing well on 0.75 mg/kg of ustekinumab at 0, 4, then Q12 weeks

Citations
