

# Congenital Anonychia

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## Disclosures

- ⦿ No financial disclosures
- ⦿ No conflicts of interest

## Case Presentation

- ⦿ History of Present Illness
  - 2 yo African American female
  - Pigmented lesion on right sole
- ⦿ Past Medical History
  - Full term infant
  - No significant medical problems
  - Meeting all milestones

## Case Presentation

- ⦿ Family History
  - Non-contributory
- ⦿ Social History
  - Lives with parents at home
  - Parents are unrelated
  - Uneventful pregnancy

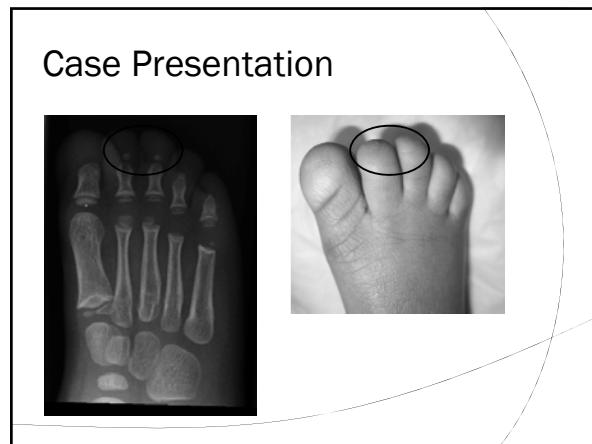
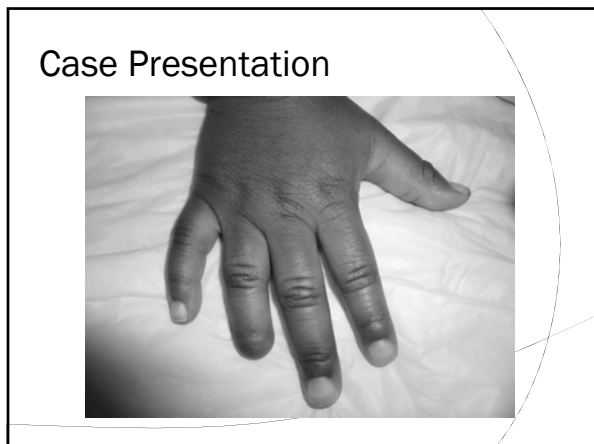
## Case Presentation

- ⦿ Physical Exam
  - Healthy appearing 2 yo female
  - 6 mm black crescentic macule on right sole



## Case Presentation





- ### Differential Diagnosis
- Nail-Patella Syndrome
  - COIF: Congenital Onychodysplasia of the Index Finger
  - Coffin-Siris Syndrome
  - Brachydactyly
  - Cooks Syndrome

### Differential Diagnosis

Table 1 Differential diagnosis of syndromes featuring anonychia and hyponychia

Syndrome	OMIM no.	Clinical features
Brachydactyly type B	113000	Hypoplasia or absence of distal phalanges DII-DV of hands, fingernail hypoplasia DII-V, lesser affected toes
Brachydactyly type A5	112900	Absence of middle phalanges and distal phalanges of hands and feet, fingernail dysplasia DII-V
Cooks syndrome	106995	Hypoplasia/Absence of DII-DV of hands and feet, hypoplasia of fingernails DI-III, anonychia DIV-V, toenail aplasia

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### Cooks Syndrome

**CLINICAL GENETICS** An International Journal of Genetics, Molecular and Personalized Medicine

**A new nail dysplasia syndrome with onychonychia and absence and/or hypoplasia of distal phalanges**

Roberta Goldshlag Cooks, Marjorie Hertz, Marissa Bat Miriam Katznelson, Richard M. Goodman<sup>†</sup>

Nevin, NC, Thomas, PS, Eady, DJ, Shepherd, C. Anonychia and absence/hypoplasia of distal phalanges (Cooks Syndrome): report of a second family. J Med Genetics 1995;32:638-641.

### Genetic Association

**Duplications of noncoding elements 5' of SOX9 are associated with brachydactyly-anonychia**

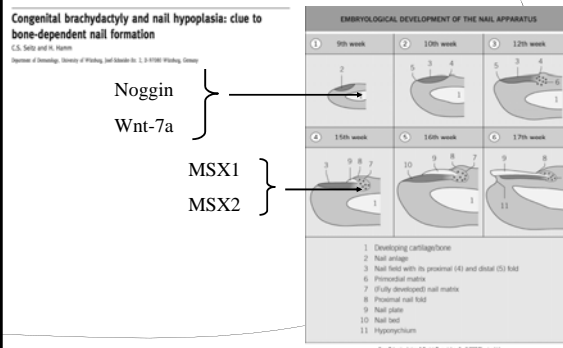
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Kurth, I, Klopocki, E, Stricker, S, van Oosterwijk, J, Vaneck, S, Altman, J, Santos, H, et al. Duplications of noncoding elements of 5' of SOX9 are associated with brachydactyly-anonychia. Nature Genetics 2009;41:886-883.

## Further Work-Up

- Consult to Genetics
  - Agreed that appearance consistent with Cooks syndrome
  - Karyotype analysis recommended
  - Patient karyotype normal

## Nail Development



## Conclusions

- Anonychia is rare, especially when it involves multiple digits
- Need to establish if nail changes are congenital or acquired
- Important to evaluate bone formation when assessing a patient with hyponychia or anonychia
- Further evaluation may be warranted including imaging, genetics consultation

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