

Hypopigmentation in a Child

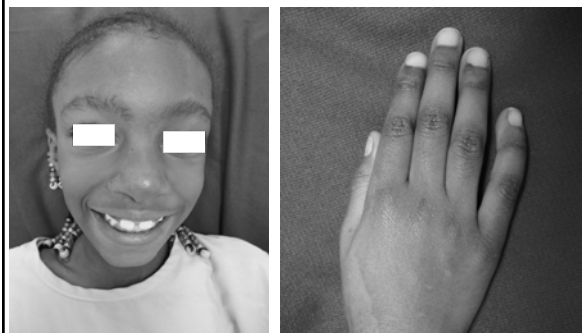
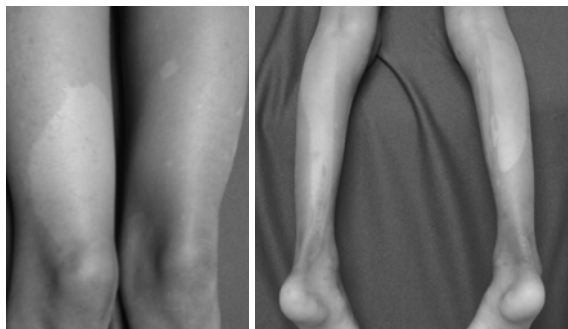
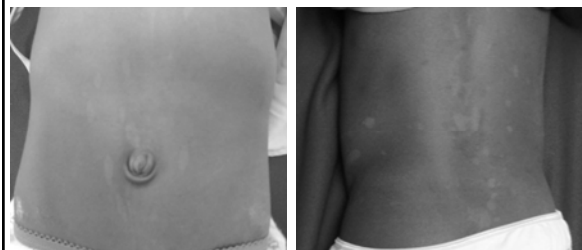
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Disclosures

- I do not have any financial relationship to disclose

Clinical Presentation

- HPI:
 - 6 yo AAF with congenital hypopigmented macules on trunk and extremities
- PMH:
 - Full term vaginal delivery
 - Meeting developmental milestones
 - Bilateral strabismus
 - Advanced bone age
 - Premature adrenarche
- FHx:
 - Uncle with strabismus



Differential Diagnosis

- Hypomelanosis of Ito
- Incontinentia Pigmenti
- Segmental Vitiligo
- Phylloid Hypomelanosis

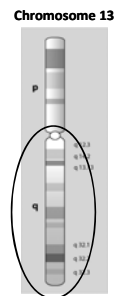
Phylloid Hypomelanosis

- 8 cases have been reported, all girls
- Greek *phylloid* = leaf-like
- Distinct syndrome with pigmentary mosaicism
 - Round or oval macules
 - Reminiscent of floral ornaments

Associated Findings

- | | |
|---|---|
| <ul style="list-style-type: none"> ■ Cerebral defects <ul style="list-style-type: none"> ■ Mental deficiency ■ Seizures ■ Ocular defects <ul style="list-style-type: none"> ■ Strabismus ■ Coloboma | <ul style="list-style-type: none"> ■ Skeletal defects <ul style="list-style-type: none"> ■ Syndactyly ■ Clinodactyly ■ Scoliosis ■ Malposition of teeth |
|---|---|

Cytogenetics

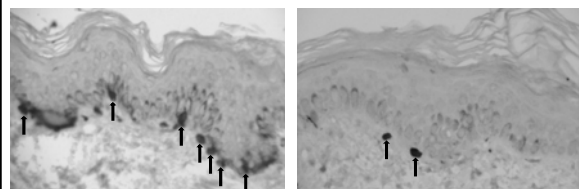


- Mosaic trisomy 13
 - 2 genetically distinct cell lines
 - 13q region

Diagnosis

- Punch bx of lesional and "normal" skin
- Immunohistochemistry for melanocytes
 - Cytogenetic studies:
 - Karyotype
 - FISH

Melanocyte Immunohistochemistry



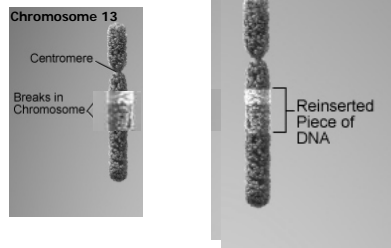
"Normal" Skin

Lesional Skin

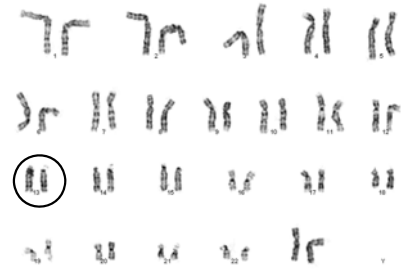
Cytogenetics Results

2 Cell lines in lesional and "normal" skin:

- 1st - 46,XX
- 2nd - trisomy 13 with paracentric inversion



Normal Cell Line



46,XX

Trisomic 13 Cell Line



47,XX,+13,inv(13)(q14q32)

Next Steps

- Peripheral blood lymphocyte analysis
- PCP, dentist and ophthalmologist
- Genetic counseling

Future Directions

- Why only in females?
- Explore mechanism for hypopigmentation
- Pigmentary genes on chromosome 13:
 - *EDNRB* and *EFNB2* – melanoblast migration
 - *DCT* – melanosomal enzyme

Summary

- Phylloid hypomelanosis is a distinct clinical entity of cutaneous findings with CNS, ocular and skeletal defects
- Our case provides additional evidence that mosaic trisomy 13 leads to phylloid hypomelanosis

References

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