Epidermal Nevus with Extensive Cutaneous Involvement

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INTRODUCTION

- Epidermal nevi (EN) are benign congenital skin lesions derived from a postzygotic mutation in a subset of pluripotential embryonic cells (mosaicism).
- The lesions tend to arrange in a whirlwind pattern representing the migration of the pluripotent cells, known as lines of Blaschko.
- The distribution and extent of EN varies greatly ranging from a single linear lesion to systemic involvement.
- More extensive lesions are highly associated with musculoskeletal and nervous system abnormalities, making up what is known as Epidermal Nevus Syndrome.
- Not only do the extent of the lesions vary greatly, but so do the underlying genetic mutations demonstrating the difficulties in defining a clear phenotype-genotype model.
- These mutations include *FGFR3*, *PIK3CA*, and *HRAS* [1,2,3,4,5]

AIM

- To help bridge the genotype-phenotype in EN, we report an atypical case of EN with pathologic and genetic analyses.

CASE HISTORY:

- A 6-month-old female presented with hyperpigmented linear and whirlwind patterned flat patches and macules that followed the lines of Blaschko on the lower face, neck, trunk, buttocks/groin, and extremities (Figure 1).
- There was no overlying erythema, blisters, erosions, or thickening of the areas.
- The lesions were present since birth and non-changing. The patient was found to have hip dysplasia requiring bracing.
- The patient had no intraoral manifestations, ocular defects, or developmental abnormalities.
- There was no family history of any dermatologic, neurologic, or skeletal abnormalities.

WORK UP:

- A punch biopsy of affected skin demonstrated mild epidermal papillomatosis, acanthosis, hyperpigmentation, and thickening of the rete ridges (Figure 2 & 3).
- A biopsy of the nonaffected neighboring skin showed no abnormalities.
- Both specimens were sent for whole-exome sequencing (results pending).

DISCUSSION:

- The clinical and pathologic findings were most consistent with epidermal nevi.
- Most common histological patterns of EN have been reported as: hyperkeratosis, papillomatosis, and acanthosis with elongation of rete ridges[6], all of which our patient had.
- EN syndrome was initially suspected as the patient had extensive lesions, but no systemic abnormalities were found.
- The differential diagnosis for EN includes pigmentary mosaicism and incontintentia pigmenti, since these can also appear as hyper/hypo pigmented lesions in lines of Blanchko, but histologic findings were inconsistent with this.

Figure 1: Patient with hyperpigmented whirlwind patterned flat patches and macules following the lines of Blaschko on the back.

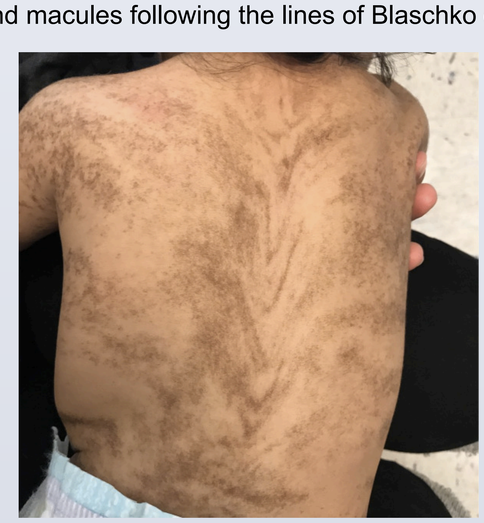


Figure 2: Biopsy of patient's affected skin showing mild epidermal papillomatosis, acanthosis, basal hyperpigmentation.

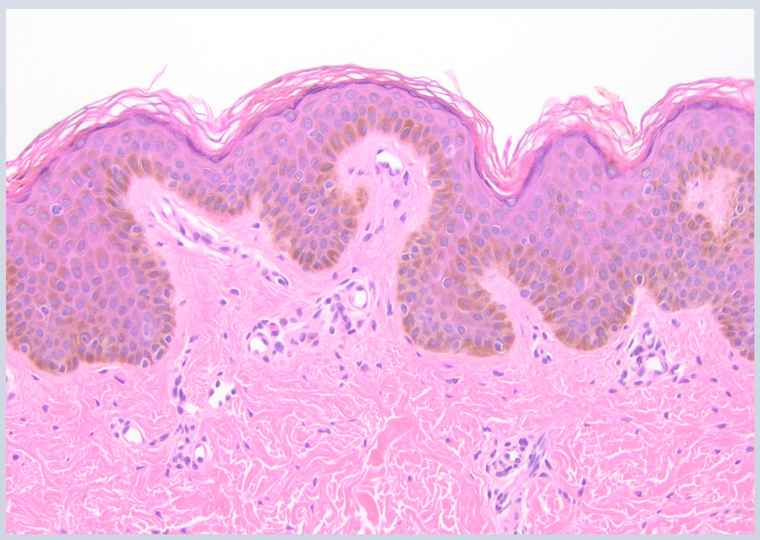
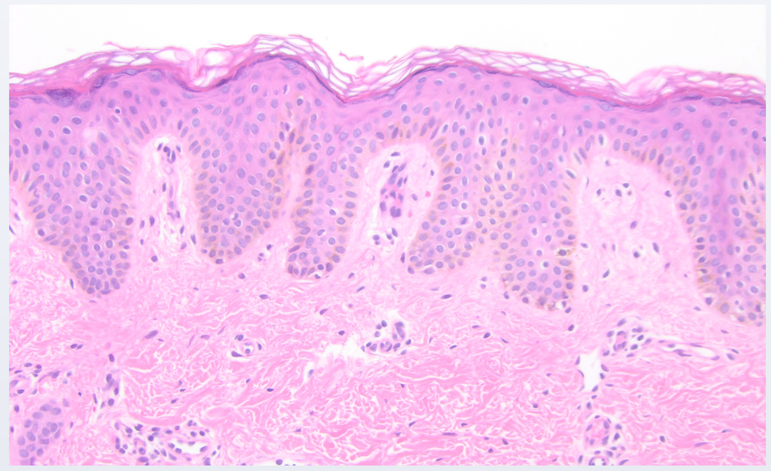


Figure 3: Biopsy of patient's affected skin showing elongation of rete ridges and papillomatosis.



CONCLUSIONS

- This case report is an example of extensive EN without systemic abnormalities.
- The phenotypic variability seen in EN may be due to the variable genetic mutations

ACKNOWLEDGEMENTS

- We are grateful for the patient and her family for participating in this study.
- The study was approved by UC Davis institutional review board (925818).

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