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

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 ridges6, 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 incontinentia pigmenti, since these can also appear as hyper/hypo pigmented lesions in lines of Blaschko, but histologic findings were inconsistent with this.

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).

REFERENCES