Intraoperative Dermoscopy for Identification of Early Basal Cell Carcinomas in Basal Cell Nevus Syndrome
Disclosures

• I have no industry related, financial, or other disclosures
Goals

• Discuss the clinical characteristics of Nevoid Basal Cell Carcinoma Syndrome

• Identify the dermascopic features of basal cells found in syndromic patients

• Discuss a highly effective and well tolerated treatment modality
  ▫ The CO2 Laser
Gorlin’s Syndrome

- **Nevoid Basal Cell Carcinoma Syndrome (NBCCS)**
  - Rare autosomal dominant mutation of PTCH gene on C-some 9q
  - Prevalence is approximately 1/100,000

- **Main Cutaneous Characteristics**
  - Multiple Basal Cell Carcinomas (BCCs)
  - Palmar and plantar pits
Diagnostic Criteria (2 Major, 1 Minor)

- **Major Criteria**
  - **Cutaneous**
    - Over 5 BCCs in a lifetime or one before age 30
    - Palmar and plantar pits
  - **Radiographic**
    - Lamellar calcification of the falx cerebri
    - Jaw keratocysts or polyostotic bone cyst
  - **Historical**
    - First degree relative with NBCCS
  - **Controversial**
    - Medulloblastoma
Diagnostic Criteria

• Minor Criteria
  ▫ Macrocephaly
  ▫ Cleft lip and/or palate
  ▫ Vertebral and/or rib abnormalities
  ▫ Polydactyly
  ▫ Ovarian and/or cardiac fibromas
  ▫ Ocular abnormalities
Variable Expressivity

105 people aged 4 months to 87 years

• BCC in 80% Whites and 38% African Americans
• Palmar pits in 80%
• Jaw cysts in 74%
• Facial dysmorphism in 55%
• Radiographic changes in up to 65%
  ▪ Falx cerebri calcification
  ▪ Bifid ribs
  ▪ Hemi- or fused vertebrae
  ▪ Flame shaped lucencies of the phalanges, metacarpal, and carpal bones
BCC  |  Palmar Pits  |  Jaw Cysts  |  Facial Dysmorphism
Clinical Challenges for Dermatologists

• Basal Cell Carcinomas (BCCs)
  ▫ Multiple
  ▫ Early
  ▫ Can be difficult to identify

• Treatment
  ▫ Safe
  ▫ Effective
  ▫ Well tolerated
  ▫ Efficient
  ▫ Cosmetically acceptable
Dermatoscopic findings in BCC

• Gross Clinical BCC Types
  ▫ Nevoid
    • Pigmented
      • Additional subtypes
        ▫ Acrochordon-like
        ▫ Hemangioma-like
  ▫ Featureless (very small BCCs)
  ▫ Typical BCC (more common with increasing age)
Nevoid BCC

- **Always present**
  - Pigment without a defined network
- **May be present**
  - Brown-grey dots and globules
  - Telangiectasia (usually lesions over 3mm)
  - Leaf-like areas (larger lesions)
  - Smooth and pearly surface
Featureless BCCs

- Very small
- Round
- Skin colored papules
- May be polypoid
Case Presentation

• 1-year-old boy
  ▫ Father with history of NBCCS
• Findings
  ▫ Macrocephaly with frontal bossing
  ▫ Multiple fused vertebrae
• By age 3
  ▫ Palmar pit on the left palm
  ▫ Several skin-colored papules with brown globules noted via dermoscopy on his occipital scalp and left eyelid
• Over the next several years, he developed over 100 BCCs
Treatment

• Treatment goals for our patient
  ▫ Safe
  ▫ Effective
  ▫ Well tolerated
  ▫ Efficient
  ▫ Cosmetically acceptable
Treatment Options for BCC in Gorlin’s Syndrome

- **Surgical**
  - CO2 Laser
  - Cryosurgery
  - Excision
  - Mohs

- **Topical**
  - Imiquimod
  - 5-FU
  - PDT

- **Targeted**
  - Vismodegib
CO2 laser

- Study by Nouri, et al.
  - Patient with phototype IV and BCNS
  - 100 BCCs on a patient’s trunk, 20 on the neck, and 2 on the face (all 1-4mm in size) identified

  - Treated with CO2 laser
    - Endpoint = skin yellowing
    - 500 mJ, 5 W, 3 mm spot size, three to four passes

  - 1 month later 6 random sites chosen for post-op Mohs sections
    - No remaining malignancy identified
CO2 Laser

- **Advantages**
  - Precision treatment and decreased postop pain
  - Low recurrence (<3% of lesions smaller than 10mm)
  - Rapid tx of many lesions with short healing time (about 2 wks)
  - Better cosmesis (minimal hyperpigmentation and scarring)
CO2 Laser

• When is it not appropriate...
  ▫ BCCs larger than 10mm
  ▫ Ulcerative, ill-defined, or very thick BCCs
Intraoperative Dermoscopy Followed by CO2 Laser Ablation

- Phoenix Children’s Hospital
  - Harper Price, MD and Ronald Hanson, MD et al.

- Patient is seen in clinic for skin check
  - BCCs identified by dermoscopy without biopsy
    - Exceptions
      - Diagnostic uncertainty
      - Larger than a few millimeters

- Patient is scheduled for CO2 laser surgery
Intraoperative

- Patient arrives at facility and undergoes induction with general anesthesia
- Dermoscopy again used intraoperatively to identify BCCs
Treatment

- CO₂ laser
  - 10,600nm, 3-5 W in a continuous fashion
  - Each lesion is treated with one to four focal passes
  - Destruction noted by the endpoint of dermal tightening and slight crust formation
Post-treatment

- Usually complete recovery is in 1-2 weeks
- Slight hypopigmentation with little if any scarring
- Recurrence does occur occasionally
Patient Satisfaction

- Cosmetic results preferred over punch excision
- Appreciate the ability to treat multiple sites in a single session
- Report minimal discomfort
Conclusions

• Basal Cell Nevus Syndrome is a disease with both diagnostic and therapeutic challenges
• Cutaneous Clues
  ▫ Early BCCs
  ▫ Palmar pits
  ▫ Facial dysmorphism
• CO2 laser
  ▫ Effective
  ▫ Efficient
  ▫ Safe
  ▫ Well tolerated
  ▫ Cosmetically acceptable
References