ECRONICON OPEN ACCESS

EC DENTAL SCIENCE
Opinion

Integumentary Transmutation - Nevoid Basal Cell Carcinoma Syndrome

Anubha Bajaj*

Department of Histopathology, Panjab University, A.B. Diagnostics, India

***Corresponding Author**: Anubha Bajaj, Department of Histopathology, Panjab University, A.B. Diagnostics, India.

Received: August 29, 2022; **Published:** September 23, 2022

Nevoid basal cell carcinoma syndrome is an inherited, multisystem disorder engendered on account of germline mutations within human homolog of patched (PTCH) gene. The condition exhibits a predilection towards soft tissue overgrowth, configuration of neoplasms and skeletal anomalies.

Additionally designated as basal cell nevus syndrome, Gorlin syndrome, Gorlin-Goltz syndrome, basal cell carcinoma nevus syndrome, fifth phakomatosis or bifid-rib basal cell nevus syndrome, nevoid basal cell carcinoma syndrome is commonly associated with a de novo genetic mutation [1,2].

Nevoid basal cell carcinoma syndrome enunciates genomic mutations within several genes of sonic hedgehog signalling pathway.

Sporadic instances are associated with dual somatic "hits" within a singular cell. Familial nevoid basal cell carcinoma syndrome exemplifies singular somatic "hit" along with inheritance of singular defective allele.

Loss of functional mutations within tumour suppressor gene PTCH1 ensures premature termination of PTCH protein, a feature which implicates multiple, diverse organs and systems [1,2].

The autosomal dominant condition demonstrates enhanced penetrance and variable expressivity. Genomic mutations within PTCH1 gene situated upon chromosome 9q22 are commonly encountered. Infrequently, PTCH2 gene situated upon chromosome 1p32 or SUFU gene situated upon chromosome 10q24-q25 are incriminated [1,2].

Nevoid basal cell carcinoma syndrome is associated with variable clinical representation and consequently delayed disease discernment. Manifestations such as odontogenic keratocysts confined to head, neck, face or jaw, medulloblastomas, ovarian fibromas, basal cell carcinomas, palmar or plantar pitting, rib anomalies, polydactyly of hands or feet, hallux valgus, pectus excavatum or pectus carinatum, syndactyly of second and third fingers, cardiac fibromas, strabismus, hypertelorism or congenital cataracts are predominant [1,2].

Nevoid basal cell carcinoma syndrome is associated with occurrence of basal cell carcinoma which appears as a prominent feature within Gorlin syndrome, Bazex-Dupré-Christol syndrome, Rombo syndrome or Xeroderma pigmentosum [1,2].

Basal cell carcinoma is a common feature of geno-dermatoses such as Bloom syndrome, Brooke-Spiegler syndrome, cartilage-hair hypoplasia, Cowden syndrome, epidermodysplasia verruciformis, exposure to arsenic and disorders of melanin biosynthesis as oculocutaneous albinism or Hermansky-Pudlak syndrome, Muir-Torre syndrome, Rothmund-Thomson syndrome, Schöpf-Schulz-Passarge syndrome or Werner syndrome [1,2].

Nevoid basal cell carcinoma syndrome requires segregation from conditions such as Bazex syndrome, fibrous papule of face, melanocytic nevi, milia, pseudo-hypoparathyroidism, seborrheic keratosis or unilateral nevoid basal cell carcinoma syndrome with comedones [3,4].

Nevoid basal cell carcinoma syndrome can be appropriately discerned with pertinent criteria which necessitate screening for the condition as occurrence of odontogenic keratocysts or basal cell carcinoma in subjects < 20 years, multiple basal cell carcinomas in subjects > 20 years, palmar or plantar pits, bilamellar calcification of falx cerebri, bifid, fused or splayed ribs, first-degree relative with nevoid basal cell carcinoma syndrome and emergence of medulloblastoma with desmoplastic histological features along with manifestation of aforesaid criteria [3,4].

Minor diagnostic criterion include manifestations such as enhanced head circumference, congenital malformations as frontal bossing, coarse facies, cleft lip/palate, moderate or severe hypertelorism, skeletal anomalies as Sprengel deformity, marked pectus deformity, marked syndactyly of digits, radiological anomalies as bridging of sella turcica, hemivertebrae, fusion or elongation of vertebral bodies, modelling defects of hands or feet or flame-shaped radiolucency of hands or feet and occurrence of ovarian and cardiac fibromas [3,4].

Evaluation of genetic mutations and sequence analysis of PTCH1 gene is optimal and recommended in subjects requiring prenatal investigation of familial mutations and predictive assessment or confirmation of individuals with partial expression of diagnostic criterion [3,4].

Magnetic resonance imaging (MRI) or computerized tomography (CT) enunciates features such as bilateral hyperplasia of mandibular coronoid processes, odontogenic keratocysts, mandibular prognathism, calcification of falx cerebri, medulloblastoma, fused vertebrae, kyphosis, lumbarization of sacrum, scoliosis, Marfanoid habitus, polydactyly, short metacarpals, flame-shaped radiolucency of hands or feet or spontaneous fractures [3,4].

Echocardiography is relevant with emergence or suspicion of cardiac fibromas [3,4].

Ultrasonography of pelvis is optimal for discerning ovarian fibroma. Premature morbidity and mortality is associated with nevoid basal cell carcinoma syndrome on account of concurrent cutaneous carcinomas and accompanying neoplasms in spite of adoption of extensive therapeutic strategies [3,4].

Cogent therapy is comprised of a multidisciplinary approach with appropriate evaluation of cutaneous, genetic, paediatric, gynaecologic, cardiac, oral and maxillofacial anomalies [3,4].

Pertinent surveillance and treatment of disease associated manifestations is beneficial [3,4].

Benign and malignant neoplasms are managed with surgical intervention or feature-specific pharmaceutical agents [3,4].

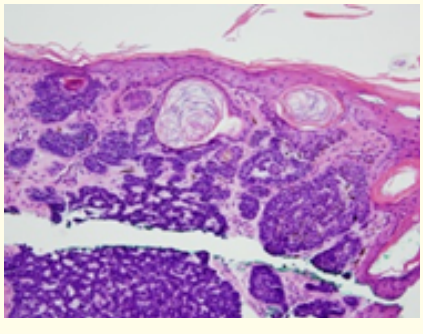

Figure 1: *Basal cell carcinoma delineating lobules of basaloid epithelial cells with peripheral palisading and enveloping fibromyxoid stroma with stromal retraction of tumour lobules. Superimposed stratified squamous epithelium adheres to subjacent tumour nests and displays mild acanthosis with hyperkeratosis [5].*

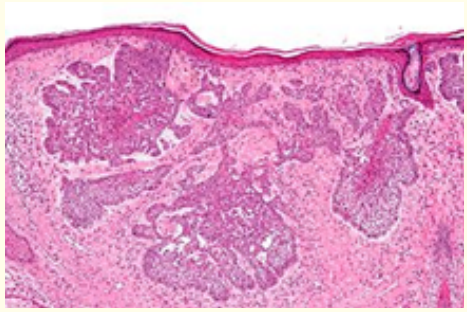

Figure 2: *Basal cell carcinoma exhibiting aggregates of basaloid epithelial cells with peripheral palisading surrounded by fibromyxoid stroma and clefts between tumour lobules and stroma. Tumour nests are adherent to superimposed epidermal layer which exemplifies mild acanthosis with hyperkeratosis [6].*

Bibliography

1. Spiker AM., *et al*. "Gorlin Syndrome". Stat Pearls International, Treasure Island, Florida (2022).
2. Hazemann G., *et al*. "Histopathological study of basaloid follicular hamartoma". *Annales de Dermatologie et de Vénéréologie* 146.3 (2019): 181-191.
3. Qiu F., *et al*. "A case of nevoid basal cell carcinoma syndrome dominated by facial basal cell carcinoma". *Zhong Nan Da Xue Xue Bao Yi Xue Ban* 47.3 (2022): 384-389.
4. Kim CS and Na YC. "Basal cell nevus syndrome with excessive basal cell carcinomas". *Archives of Craniofacial Surgery* 22.2 (2021): 122-125.
5. Image 1 Courtesy: eScholarship.com.
6. Image 2 Courtesy: Libre pathology.

Volume 21 Issue 10 October 2022