To report a case of a patient with Gorlin Syndrome – its clinical presentation, diagnostic criteria, and holistic management.

OBJECTIVES

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MATERIALS AND METHODS

RESULTS

This section briefly describes the history, physical examination findings, and the surgical management done for the patient.

DISCUSSION

GORLIN SYNDROME

First described in 1894, and more clearly defined in 1960 by Gorlin and Goltz

Rare autosomal dominant, tumor-predisposing disorder by PTCH1/SUFU gene mutations

Estimated prevalence can be low as 1 in 164,000, but may be higher since mild conditions are unrecognized

Only 2 cases have been published in the Philippines

MAJOR CRITERIA

Early development (around 20 years of age) of multiple basal cell carcinomas

1st degree relative with Nevoid BCC

≥ 2 visible palmar and/or plantar pits

Lamellar calcification of the falx cerebri

Jaw keratocysts on imaging

MINOR CRITERIA

Skeletal anomalies (i.e. polydactyly or a splayed vertebra or rib)

Ocular anomalies (i.e. cataract, developmental defects, etc.)

Lymphomesenteric, pleural, or ovarian cysts and fibroma

Congenital anomalies (i.e. cleft lip and/or palate, macrocephaly, and medulloblastoma)

The patient has 3 major and 3 minor features – fulfilling the Diagnostic Criteria of Gorlin Syndrome of at least 2 major, or 1 major and 2 minor features

TREATMENT

Surgical excision

PREVENTION

Avoidance of sun exposure or Vitamin A supplementation

SURVEILLANCE

Annual chest/jaw x-ray and skin exam, including relatives

GENETIC COUNSELLING

75% of patients have an affected parent, and 25% of offspring can have the condition

CONCLUSION

Gorlin Syndrome, although not usually life-threatening, is an aggressive and recurrent disease that leads to gross disfigurement.

The ability to identify this syndrome, empathetic patient education, and active surveillance remain as the physician’s best approach in treatment and management.

REFERENCES