Cylindromatosis, Familial

Alternative Names
CYLD
Ancell-Spiegler Cylindromas
Turban Tumor Syndrome
Cylindromas, Dermal Eccrine
Turban Tumors

Record Category
Disease phenotype

WHO-ICD
Neoplasms > Benign neoplasms

Incidence per 100,000 Live Births
Unknown

OMIM Number
132700

Mode of Inheritance
Autosomal dominant

Gene Map Locus
16q12-q13

Description
Familial cylindomatosis is an autosomally inherited genetic condition, characterized by numerous benign tumors of skin adnexa, principally on the head and neck. The tumors arise from the hair follicles, sweat glands, and sebaceous glands, and exist as circumscribed, non-encapsulated, dermal forms without any attachment to the epidermis. In addition, the tumors are made up of numerous oval and angulated subunits, arranged in an interlocking pattern. In fact, these angulated subunits are what differentiate the tumor from spiradenomas. Other neoplasms that share clinical findings with cylindromatosis include Brooke-Speigler Syndrome, and Multiple Familial Trichoepithelioma. The subunits are surrounded by PAS positive, glassy, basement membrane.

Cylindromatosis usually affects individuals in their second or third decade of life, and shows a female preponderance. Malignant transformation is very rare. Treatment involves surgical removal and skin grafting.

Molecular Genetics
The gene responsible for familial cylindromatosis is the CLYD gene on chromosome 16q12-q13. The normal function of the CLYD gene is complex and diverse. CLYD has been shown to interact with various members of the pro-apoptotic NF-Kappa B signaling pathway, including TRAF-2 (TNF receptor Associated factor 2), TRIP (TRAF Interacting Protein), and IKK gamma/Nemo (I Kappa B Kinase, Gamma), and thereby, negatively regulating NF-Kappa B activation. Thus, loss of action of CLYD leads to a loss of apoptotis control, ensuing in cell proliferation. Recently, aspirin has been shown to counteract the negative effects of CLYD inactivation and induce apoptosis, and a treatment with aspirin is being considered for cylindromatosis.

Epidemiology in the Arab World
Bahrain
Al-Arrayed and Vaidya (1981) described two rare cases of adenocystic carcinoma arising from the lacrimal glands. The first case was that of a 16-year old Bahraini female, who presented with slight discomfort and redness in the left eye. She was seen to have a slight proptosis in the left eye, and a firm mass was felt in the upper outer quadrant upon palpitation, which was confirmed by the X-ray studies. The mass was excised out by surgery, and the biopsy result was that of adenocystic carcinoma of the lacrimal glands. The tumor was encapsulated and showed a pattern of cords and small columns of basal like cells. The cystic spaces surrounding the cells were filled with mucin. Follow-up after the biopsy did not show any local recurrence or distal metastasis, and exenteration was performed three months later.

One year after the excision, however, a local recurrence was suspected, in the form of a firm nodular swelling on the outer part of the orbital margin. The patient was re-operated upon, and the mass excised. Histopathological tests again confirmed the tumor to be adenocystic carcinoma. At the time of the report, two and a half years after the surgery, the patient had had no further recurrences. The second case was that of a 26-year old Bahraini female. She presented with pain and protrusion of the left eye ball, and left sided headache of one year duration. Scan showed diffused swelling of the supra-orbital bone with stippling. The swelling was considered to be a pseudotumor, and the patient was put on systemic antibiotics and steroids. However, two days later she complained of increased pain, proptosis, and headache. A palpable mass was noticed on the upper quadrant of the orbit. Incision biopsy revealed adenocystic carcinoma of the lacrimal gland. The tumor was found to be composed of cells set in abundant fibrotic stroma with aciner foundation and occasional cysts. The tumor then grew rapidly, and although exenteration was attempted, it could not be achieved due to the massive infiltration of the tumor into the soft tissues and bones of the skull. The patient was sent to Kuwait for radiotherapy. Seven months later, she was re-admitted to the hospital, and skull X-ray showed advanced spread of the tumor with marked bony metastasis of the right orbit and skull bones. A week later, she fell from the hospital bed, and succumbed to the resulting severe epistaxis from the fragile tumor extending in the sinuses.

References

Al-Arrayed H, Vaidya PL. Behavioural patterns of adenocystic carcinoma of lacrimal glands. Bahrain Med Bull. 1981; 3(3):91-6.

Related CTGA Records

N/A

External Links

http://www.emedicine.com/derm/topic94.htm
http://www.medterms.com/script/main/art.asp?articlekey=26101
http://www.signaling-gateway.org/update/updates/200310/nrc1202.html

Contributors

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