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着色性干皮病：二家系三例报告

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提 要

本文报道着色性干皮病二家系三例，双亲均近亲婚姻，为常染色体隐性遗传。1例15岁时右眼内眦区发生鳞状细胞癌，放射治疗愈，一年后复发。简略复习讨论了病因、临床表现和防治。

着色性干皮病是一罕见皮肤病，又称着色性皮萎缩或萎缩性着色血管瘤，以日照后皮肤现雀斑样色素斑、晚期癌变及常并发眼征为主要特征，一般视为癌前期病。Kaposi（1879）首次命名[1]，国内已有报道[2][3]。现简略报告二家系三例于下。

家系及病例报告

家系一（图1）：17岁Ⅳ4，农。11岁面部出现雀斑，1980年9月（15岁）右眼内眦区长包块并

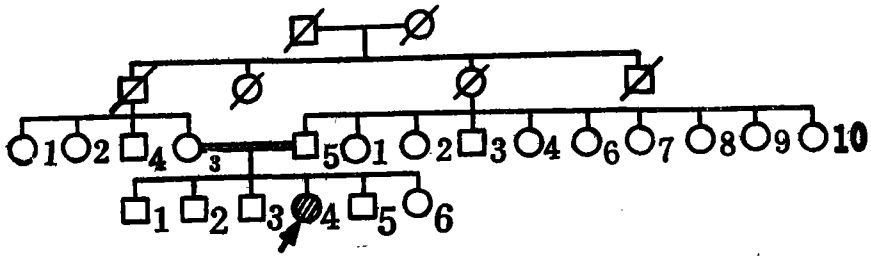


图1 着色性干皮病家系一 Ⅲ3、4—姑舅表兄妹婚 Ⅳ4—17岁先证者

溃疡，病检诊断鳞状细胞癌，X线照射后愈。1年后下睑又糜烂。1982年2月18日检查：右眼下睑缘红肿、秃睫、睑球粘连，球结膜充血肿胀，内下部尤甚，内眦区有一约1厘米直径的近园形浅溃疡，底和边缘不平，附脓性分泌物，眼球外上移位，角膜、前房及眼底未见异常。再8月后，鳞癌溃疡直径扩至4厘米，累及内半角膜，左眼正常。面、睑、颈背及双上肢伸侧散布褐色斑点，间少许脱色斑点。双亲为姑舅表兄妹婚。

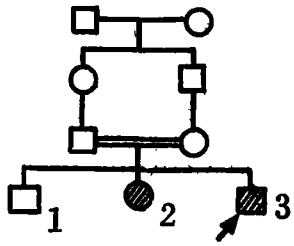


图2 着色性干皮病家系二
Ⅲ1、2——姨表兄妹婚
Ⅳ2——3岁
Ⅳ3——半岁先证者

家系二（图2）：半岁Ⅳ3，2月前面部始现雀斑，近日羞明并双眼充血。1957年5月21日检查：面、睑部散布很多褐色及白色斑点；结膜充血、血管充盈纡曲、少量出血并色素沉着；角膜浑浊并色素沉着，荧光素（一）；前房正常；眼底不见。双亲为姨表兄妹婚。

Ⅳ2 3岁，亦4月发病，病情同Ⅳ3。

讨 论

本病为常染色体隐性遗传，双亲多近亲婚。近年探明是一种生化缺陷遗传病，主要是内切核酸酶缺乏[4]，对紫外线损害的成纤维细胞DNA不能进行正常的切除修复[5][6]。

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本病成纤维细胞DNA的修复率只有常人的20%或缺如。故可能隐性致病基因决定成纤维细胞DNA的修复功能缺陷，而日照为发病诱因。McCormick等发现培养的着色性干皮病细胞也对多环芳烃及其它致癌因子如黄曲毒素等较敏感，突变率较高[8]，本病可能在许多因素作用下诱发。

本病多在生后1年内的春夏季节发病，好发于暴露部皮肤如面、颈和四肢等部位。先出现红斑、水疱，接着色素斑和干燥萎缩，最后多发性癌变。眼部常累睑和角、结膜：色素沉着，充血肥厚，睑球粘连及睑内外翻，最后赘疣形成和癌变。予后不良，多因癌变(主要是鳞状细胞癌、基底细胞癌和恶性黑色素瘤[4]) 或免疫缺陷致呼吸系统感染[7]于成年以前死亡[2]。治疗困难，早期可切除加放射治疗，5-荧光-尿嘧啶等细胞毒药物可能有效。预防：应注意避免日照，不要接近诸如燃煤、吸烟及毛发染料等，并勿食用黄曲菌感染的食物，禁近亲婚姻。

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Xeroderma Pigmentosum
——A Report of Three Cases from Two Families

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Xeroderma pigmentosum is a rare inheritable skin disease with autosomal recessive trait often resulted from close relative marriage. The primary defect seems to be a deficiency of endonuclease, the lack of which interferes the normal repairment and cleavage of the damaged DNA of the fibroblasts caused by ultraviolet

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racteristics and diagnostic criteria and assessable prognosis and to accumulate the clinical material for research purposes.

Thirty-seven patients out of the 50 have been followed up for years, the longest up to 19 years. 14 patients(37.8%)developed local recurrences. The five and ten year survival rates are 62.2% and 37.8% respectively.

The differential diagnosis, histochemistry, histogenesis, and prognosis of ACC were discussed with a brief review of the literature concerned. Our data supports the assumption that ACC originates from the intercalated duct cells rather than from fully differentiated serous cells.

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irradiation.

Three cases of xeroderma pigmentosum from two families were presented. One of the patients was a 17-year-old peasant girl, the fourth daughter of a couple who were first cousins before marriage. A squamous cell carcinoma was found in her right medial canthus when she was 15. The tumor disappeared after x-ray treatment but recurred one year after. The cancerous ulcer was 4 cm in diameter and involved the internal half of the right cornea on admission. In addition multiple brownish spots were seen scattering all over her face, eyelids, back of the neck, and two upper extremities. The other two patients were a two-year-old girl and a three-year-old boy, the parents of whom were also first cousins before marriage.

The clinical manifestations, prevention, and treatment of this malady were reviewed and discussed.

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