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(收稿日期 2006-03-22)

(本文编辑 刘萍)

## · 病例报告 ·

## 先天性丑角脸一家系 10 例

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先证者(Ⅲ<sub>3</sub>)男,82岁。先天性颜面发育畸形:眼距宽,眼裂小,额鼻部两眼内眦间皮肤明显增厚隆起,表面平坦,肤色正常,外观颇似京剧丑角脸谱中额鼻处的白斑形状。查体:双眼内眦处可见蚕豆大小的扁平赘肉,掩藏于上眼睑下。鼻骨无塌陷,视觉、听觉及发音均无明显障碍。智力正常,身体健康,未发现其他先天性畸形。

家系调查:据患者追忆其外祖母、母亲均有类似畸形,舅舅正常,姐姐亦有类似外貌。患者女儿及外甥均有此畸形。此3人均各育有两子均一正常一畸形。第5代6人中有3人已婚育,其中2名正常者均育有1正常男孩,1畸形者育有1女儿也正常,其他3人尚未婚育。该家系共6代28人中有10例患者,男女均有,病症表现无轻重差别,均为先天性畸形,无随年龄变化趋势。家系图见图1。

讨论 根据患者临床特征和家系图谱分析,我们认为该家系符合常染色体显性遗传可能性大。常染色体显性遗传有4个遗传特点:(1)患者双亲之一必是患者;(2)双亲无病时,子女中无发病的患者。以上两点该家系均符合。(3)连续遗传。该家系前5代均连续遗传,第6代3人均正常,无患者。虽然如

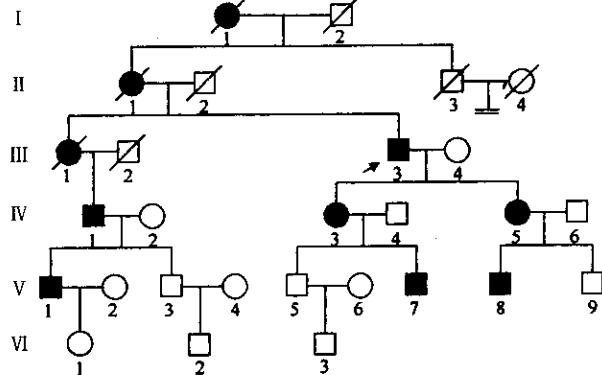


图1 先天性丑角脸遗传家系图

此,我们分析其原因(1)第5代3位患者仅1人结婚生育,其余2人尚未婚(2)由于计划生育,生育子女数减少,常染色显性遗传疾病患者子女可有1/2正常率,如其余2人生育也可能如V<sub>1</sub>一样子女正常,那么此种疾病将从此中断不会再遗传给后代。该家系第2、3代均为患者,也与子女数较少,造成偏差有关。

(收稿日期 2006-04-27)

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