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· 病例报告 ·

先天性丑角脸一家系 10 例

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先证者(Ⅲ₃)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

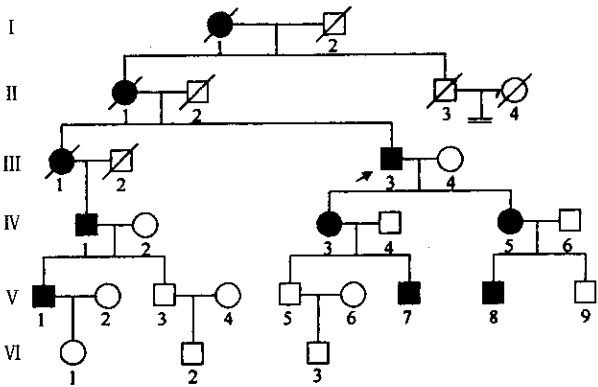


图 1 先天性丑角脸遗传家系图

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 V₁ 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。

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