

功能性面部疾病的临床诊断与治疗

潘小玲 王刚

【摘要】 功能性运动障碍是临床常见的转换障碍,累及面部者称为功能性面部疾病,影响患者社交和工作能力。目前大多数临床医师对此类疾病认识尚不充分,使疾病诊断与治疗不及时。本文综述功能性面部疾病的临床表现、电生理学特征、诊断与鉴别诊断、临床管理,以提高临床诊断与治疗水平。

【关键词】 转换障碍; 面部肌肉; 综述

Diagnosis and treatment of functional facial disorders

PAN Xiao-ling¹, WANG Gang²

¹Department of Neurology, Jinhua Municipal Central Hospital, Jinhua 321000, Zhejiang, China

²Department of Neurology, Ruijin Hospital, Shanghai Jiaotong University School of Medicine, Shanghai 200025, China

Corresponding author: WANG Gang (Email: w11424@rjh.com.cn)

【Abstract】 Functional movement disorder (FMD) is a common conversion disorder, and those who involve the face are called functional facial disorders. It seriously affects patients' social contact and work. Most clinicians have insufficient understanding of functional facial disorders, resulting in delayed diagnosis and treatment. This paper reviews the clinical features, electrophysiology, diagnosis and differential diagnosis, and treatment of functional facial disorders to improve the level of diagnosis and treatment.

【Key words】 Conversion disorder; Facial muscles; Review

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功能性运动障碍(FMD)累及面部时称为功能性面部疾病(functional facial disorders)或面部功能性运动障碍,目前尚未被临床医师充分认识。现代神经病学之父 Charcot 教授于 1887 年报告首例单侧癱症性面部痉挛患者,由此开启了功能性面部疾病临床研究之先河^[1-2];其后,英国神经病学家 Gowers 教授(现代神经病学奠基人之一)不仅详细描述了面部肌肉歇斯底里样强直性痉挛的症状与体征,并首次提出了错向舌偏斜(a wrong way tongue deviation)可以作为诊断线索^[2];之后相继有多位学者详细描述了功能性眼睑痉挛的临床特征,以及钩状舌和歇斯底里痉挛等功能性面部疾病的舌部特

征^[3-5]。然而,由于功能性面部疾病的跨学科特点使其研究进展缓慢,直至近 20 余年相关临床研究才逐渐增多^[6],笔者以功能性面部疾病[functional (psychogenic) facial disorders]、功能性眼睑痉挛[functional (psychogenic) blepharospasm]、功能性面肌痉挛[functional (psychogenic) hemifacial spasm]、功能性舌部运动障碍[functional (psychogenic) tongue movement disorders]、功能性面部运动障碍[functional (psychogenic) facial movement disorders]、功能性头部运动障碍(cranial functional movement disorder)和功能性运动障碍[functional (psychogenic) movement disorders]等关键词作为检索词,分别对美国国立医学图书馆生物医学文献数据库(PubMed)、Web of Science、荷兰医学文摘(Embase)、中国知网中国知识基础设施工程(CNKI)、万方医学数据库等中英文数据库进行检索,共检出功能性面部疾病相关文献 20 余篇,其中近 5 年文献 6 篇、5~10 年 10 篇,其余文献均发表于

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作者单位:321000 浙江省金华市中心医院神经内科(潘小玲); 200025 上海交通大学医学院附属瑞金医院神经内科(王刚)

通讯作者:王刚,Email: w11424@rjh.com.cn

10年前。目前尚无针对功能性面部疾病的大规模人群流行病学调查,根据运动障碍性疾病或肉毒毒素治疗中心的数据,功能性眼睑痉挛占全部功能性运动障碍性疾病的0.3%~20.0%,且不同医疗中心数据差异较大^[7-9]。2012年,Fasano等^[10]的多中心研究显示,约16.3%的功能性运动障碍患者同时存在面部受累症状;Jankovic教授团队历时近20年的单中心数据显示,功能性眼睑痉挛患者在2001、2011和2017年同期所有拟诊眼睑痉挛患者中的比例分别为2.4%^[11]、7.4%^[12]和9.8%^[13],而临床实践中功能性面部疾病的发病率远高于文献报道。本文拟就该病的临床表现、电生理学特征、诊断与鉴别诊断、临床管理等进行综述,以期对疾病的诊断与治疗提供参考,提高临床诊断率与治疗率。

一、临床表现

功能性面部疾病以面部肌肉过度活动为主,可单独发生亦可合并躯体运动障碍,好发于女性,各年龄阶段均可发病,高峰发病年龄约40岁^[10,14]。临床特征为突发突止,持续时间长短不一,病程中症状波动较大,可随注意力分散而减轻或终止,可与躯体疾病或精神心理疾病共病如非典型面部疼痛、偏头痛、慢性疲劳等^[15]。最常见的发作模式为眼睑、下唇、颈阔肌等不同组合的强直性痉挛,以单侧多见^[10,14]。根据面部受累部位,可以分为双侧眼睑受累、单侧下面部和(或)眼睑受累、单侧或双侧眼球受累、双侧下面部及舌肌受累、上腭受累。

1. 双侧眼睑受累 主要为功能性眼睑痉挛,表现为无眼轮匝肌痉挛的状态下,皱眉肌和降眉肌收缩致眉毛降低^[9];而器质性眼睑痉挛如梅杰综合征仅在眼轮匝肌痉挛时方才出现皱眉肌和降眉肌收缩致眉毛降低(称为Charcot征;图1a,1b)。此外,还可伴发一些器质性眼睑痉挛未见的眼部症状,如突发性视力丧失、动眼危象或眼球会聚痉挛^[16];与器质性眼睑痉挛相反,功能性眼睑痉挛通常在休息时眨眼增加^[17]。尽管患者进行分散注意力动作(如高声报数等)时症状改善有助于诊断功能性面部疾病,但是由于高声说话亦可降低器质性眼睑痉挛的严重程度和频率,故在功能性眼睑痉挛的诊断中慎用分散注意力动作^[17]。除功能性眼睑痉挛外,少数患者还可表现为无法睁眼,类似睁眼失用症,但其闭眼力度随检查者对眼睑施加外力的增加而增加^[18],是特异性诊断线索。

2. 单侧下面部和(或)眼睑受累 单侧下面部和

(或)眼睑受累是功能性面部疾病的常见表现,称为功能性面肌痉挛。单纯下面部受累表现为一侧下唇持续性侧向和(或)向下拉扯,伴下颌偏斜,称为拉唇征(lip-pulling test)^[19-20];同时合并同侧眼轮匝肌或颈阔肌过度收缩,呈假笑(smirk)^[10],因此认为,拉唇征和假笑是功能性面部疾病的诊断线索^[19-20]。单侧面肌受累应注意与面肌痉挛相鉴别,与面肌痉挛患者受累肌肉同步发作性抽搐不同^[21],大多数功能性面肌痉挛患者表现为受累肌肉非同步且持续性强直收缩^[19];面肌痉挛患者眼轮匝肌和额肌同时收缩致眉毛抬高,称为Babinski其他征(Babinski's other sign,图1c),诊断特异度达100%^[22],而功能性面肌痉挛患者闭眼侧眉毛降低、对侧眉毛抬高,称为Babinski其他征缺失^[13,19],此为特异性诊断线索(图1d,1e);此外,高达80%的面肌痉挛患者睡眠期仍有发作^[23],而功能性面肌痉挛患者通常睡眠期无发作^[10,12]。

3. 眼球受累 功能性眼动障碍的临床评估较为困难,需全面的眼科检查来触发或发现异常^[3,24]。

(1) 功能性眼球会聚痉挛(functional convergence spasm):是常见眼球受累类型^[25],表现为间歇性复视和视物模糊,尤其是集合近点固视后远距离注视时,单侧或双侧眼球仍内收,易误诊为单侧或双侧外展神经麻痹,但症状仅持续数秒即可自行缓解。(2) 功能性眼球会聚麻痹(functional convergence paralysis):临床相对少见,其特征为完全或部分眼球会聚受限,导致近距离注视时出现复视或视物模糊,患者通常主诉阅读困难^[3]。(3) 功能性凝视受限(functional gaze limitation):通常非患者主诉,而是眼科检查时发现,表现为眼动检查时无法垂直或水平眼动,并出现眼睑收缩、面部狰狞或做鬼脸等动作,甚至伴有疼痛,但视动刺激或头部被动运动时眼动正常^[3]。(4) 功能性眼震(functional nystagmus):亦为眼科检查所发现,与器质性眼震不同,功能性眼震无眼震慢相。值得注意的是,少数健康人同样可见生理性眼震,类似钟摆型眼震,亦无眼震慢相,但持续时间极短(仅2~5秒),可资与功能性眼震相鉴别。Baizabal-Carvalho和Jankovic^[26]则认为,上述缺乏眼震慢相的异常眼动应称为功能性扫视振荡(functional saccadic oscillations)。(5) 其他罕见的功能性眼动障碍:包括功能性眼球阵挛(functional opsoclonus)、功能性动眼危象(functional oculogyric crisis)等。

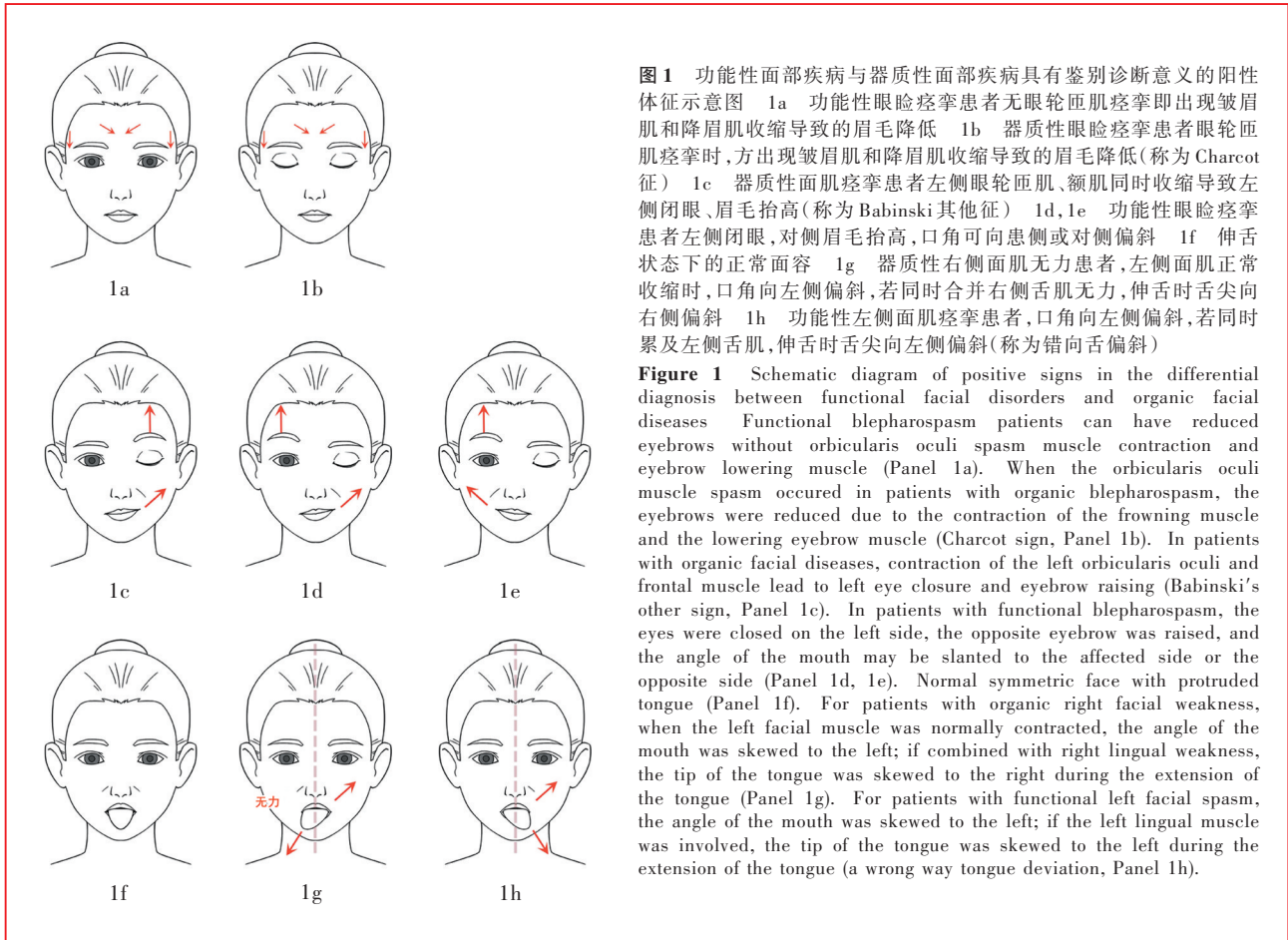


图 1 功能性面部疾病与器质性面部疾病具有鉴别诊断意义的阳性体征示意图 1a 功能性眼睑痉挛患者无眼轮匝肌痉挛即出现皱眉肌和降眉肌收缩导致的眉毛降低 1b 器质性眼睑痉挛患者眼轮匝肌痉挛时,方出现皱眉肌和降眉肌收缩导致的眉毛降低(称为 Charcot 征) 1c 器质性面肌痉挛患者左侧眼轮匝肌、额肌同时收缩导致左侧闭眼、眉毛抬高(称为 Babinski 其他征) 1d,1e 功能性眼睑痉挛患者左侧闭眼,对侧眉毛抬高,口角可向患侧或对侧偏斜 1f 伸舌状态下的正常面容 1g 器质性右侧面肌无力患者,左侧面肌正常收缩时,口角向左侧偏斜,若同时合并右侧舌肌无力,伸舌时舌尖向右侧偏斜 1h 功能性左侧面肌痉挛患者,口角向左侧偏斜,若同时累及左侧舌肌,伸舌时舌尖向左侧偏斜(称为错向舌偏斜)

Figure 1 Schematic diagram of positive signs in the differential diagnosis between functional facial disorders and organic facial diseases Functional blepharospasm patients can have reduced eyebrows without orbicularis oculi spasm muscle contraction and eyebrow lowering muscle (Panel 1a). When the orbicularis oculi muscle spasm occurred in patients with organic blepharospasm, the eyebrows were reduced due to the contraction of the frowning muscle and the lowering eyebrow muscle (Charcot sign, Panel 1b). In patients with organic facial diseases, contraction of the left orbicularis oculi and frontal muscle lead to left eye closure and eyebrow raising (Babinski's other sign, Panel 1c). In patients with functional blepharospasm, the eyes were closed on the left side, the opposite eyebrow was raised, and the angle of the mouth may be slanted to the affected side or the opposite side (Panel 1d, 1e). Normal symmetric face with protruded tongue (Panel 1f). For patients with organic right facial weakness, when the left facial muscle was normally contracted, the angle of the mouth was skewed to the left; if combined with right lingual weakness, the tip of the tongue was skewed to the right during the extension of the tongue (Panel 1g). For patients with functional left facial spasm, the angle of the mouth was skewed to the left; if the left lingual muscle was involved, the tip of the tongue was skewed to the left during the extension of the tongue (a wrong way tongue deviation, Panel 1h).

4. 双侧下面部及舌肌受累 少数功能性面部疾病患者可累及双侧下面部或双侧交替受累^[10]。舌肌强直收缩引起的舌偏斜是常见表现,通常与功能性面肌痉挛同侧,称为错向舌偏斜(图 1f~1h),是特征性诊断线索^[2]。口下颌部固定肌张力障碍相对常见,由面部外周轻微损伤所致^[27],或发生于牙科手术后数小时至数月^[20,28]。功能性舌肌震颤较为罕见,有时可合并腭肌震颤,甚至影响语言功能,表现为口吃^[29]。此外,单次问诊中功能性面部疾病患者言语障碍特征及严重程度可能随注意力分散或暗示发生明显变化^[30],如间歇性出现口吃、口齿清晰度波动性变化(时好时坏),并且可伴随一些撅嘴、眼眶周围肌肉或颈阔肌收缩等夸张动作^[31]。

5. 上腭受累 上腭震颤很少与其他部位功能性运动障碍同时出现,呈孤立性发病。上腭震颤分为原发性和继发性,继发性由 Guillain-Mollaret 三角区病变所引起,通常伴下橄榄核假性肥大^[32];原发性无明显中枢神经系统病变,其特征为软腭节律性运动,患者可听到耳内咔哒声,其病理生理学机制尚未阐明,由于绝大多数原发性上腭震颤患者对安慰

剂反应良好,故认为其可能为功能性疾病^[33-34]。上腭震颤频率、幅度和方向可变性,夹带效应或分散注意力时症状改善者,可诊断为功能性上腭震颤。

二、电生理学特征

2021 年,《功能性运动障碍的诊断与治疗中国专家共识》^[15]推荐,结合功能性面部疾病的临床特点,除采用功能性运动障碍量表、神经心理学测验量表、神经影像学等常用辅助检查外,还可选择一些特异性电生理学检查,以辅助诊断或与器质性疾病相鉴别^[35]。(1)眨眼反射:拟诊可能的功能性眼睑痉挛眨眼反射正常,而器质性眼睑痉挛眨眼反射恢复周期延长^[36]。(2)异常肌反应(AMR):亦称侧方扩散反应(LSR),是面肌痉挛的特征性肌电表现,而功能性面肌痉挛患者无异常肌反应。(3)面肌肌电图结合面部标记的视动-运动捕捉系统:用于记录患者进行注意力分散动作时、安慰剂作用下或自发性出现的不一致症状,表现为变异性显著增加,已见于功能性睁眼失用症的报道^[37]。

三、诊断与鉴别诊断

1. 诊断 功能性面部疾病的诊断主要依靠临床

病史、症状与体征,发作时完整的视频资料尤为重要,功能性运动障碍量表、神经心理学测验量表和电生理学检查可提供具有诊断价值的信息,目前主要采用 Fahh 和 Williams^[38] 制定的功能性运动障碍诊断标准。值得注意的是,功能性面部疾病的诊断主要基于阳性症状或体征,而非排除性诊断^[10,14,39],常见的阳性症状或体征主要包括^[6,40]:(1)眉毛降低。功能性双侧眼睑痉挛患者无眼轮匝肌痉挛即出现皱眉肌和降眉肌收缩导致的眉毛降低(图 1a)。(2)Babinski 其他征缺失。Babinski 其他征具有高度的诊断特异性,功能性面肌痉挛累及眼轮匝肌时,闭眼侧眉毛降低、对侧眉毛抬高,即 Babinski 其他征缺失(图 1d,1e)。(3)错向舌偏斜。功能性面肌痉挛累及舌部时,舌肌强直性痉挛,导致舌尖偏向患侧(口角上抬一侧,图 1h)。(4)拉唇征及假笑。拉唇征指功能性面部疾病累及口轮匝肌时,患侧下唇在口角处向下拉唇、偏移的现象^[19-20];可同时合并同侧眼轮匝肌和(或)颈阔肌受累,称为假笑^[13]。

2. 鉴别诊断 多种器质性神经系统疾病均有面部受累症状或体征,应注意与之鉴别。(1)面肌痉挛:表现为面部肌肉不自主抽动,一般先累及眼轮匝肌,再逐渐扩展至同侧面面部表情肌和口轮匝肌,通常单侧发病,双侧发病者极为罕见。Charcot 征(图 1b)、Babinski 其他征(图 1c)、睡眠期仍有面肌抽动以及电生理学检查显示异常肌反应等,可资与功能性面肌痉挛相鉴别。(2)Meige 综合征:常见于中老年女性,以双侧眼睑痉挛-口下颌肌张力障碍为主要表现,感觉诡计是其临床特征之一,即患者讲话、咀嚼、打哈欠、吹口哨、唱歌、敲打颈部后方时临床症状可明显缓解;Charcot 征可资与功能性眼睑痉挛相鉴别。(3)迟发性运动障碍(tardive dyskinesia):既往有明确的多巴胺受体阻断药或多巴胺耗竭药应用史,表现为重复、协调、看似有目的的口面部运动,发作刻板且症状持续存在可资与功能性面部疾病相鉴别。(4)抽动秽语综合征(TS):多见于儿童和青少年,可累及面部、颈肩部、四肢等,具有突发、快速、刻板、无目的、无节律特点,发病年龄早和显著发声异常可资与功能性面部疾病相鉴别。

四、临床管理

功能性面部疾病的治疗包括宣传教育(即疾病诊断解释)、药物治疗和非药物治疗(物理治疗和心理咨询等)。(1)宣传教育:对疾病的解释自身即具有治疗价值^[41-42]。有效的疾病解释包括肯定患者的

症状是真实存在的、给出中性诊断标签(以“功能性”替代“心因性”)、与患者一起讨论症状是如何出现的;强调常规检查正常说明无神经系统结构损害,故症状可逆;临床医师可提供长期有效的治疗管理,由此获得患者的信任并建立战胜疾病的信心。(2)药物治疗:目前尚无特效药物,主要是针对共病(焦虑、抑郁等)的对症治疗,包括选择性 5-羟色胺再摄取抑制剂(SSRI)如西酞普兰、帕罗西汀、文拉法辛等;以及特殊的诊断性治疗,如小剂量肉毒毒素肌肉注射治疗功能性面肌痉挛,数分钟内症状即显著改善^[43]。(3)物理治疗:功能性面部疾病的物理治疗更多是伴随注意力转移的运动再训练^[44],通过分散注意力、减少患者自我关注以改善症状;以及向患者展示其分散注意力时的正常面肌活动视频或既往治疗成功案例的视频,有可能具有一定疗效。最新的综述肯定经颅磁刺激(TMS)治疗功能性运动障碍的潜在前景^[45],但尚待更多研究以确定最佳刺激参数。(4)心理治疗:主要包括认知行为法(CBT)^[44]。对于准备接受心理治疗的患者而言,治疗启动时机十分重要,心理治疗开始时间太早,如疾病诊断解释阶段即予以心理治疗往往适得其反;心理治疗开始的良好时机可能是患者对疾病诊断有良好理解,并从前期治疗中开始获益之时。

综上所述,功能性面部疾病尚未得到足够重视,临床医师对其自然病程的演化还知之甚少,早期诊断、及时多学科综合治疗对改善患者生活质量具有积极意义,长期的随访管理十分必要。

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利益冲突 无

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