

# 非综合征性唇腭裂的皮纹学研究进展

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[摘要] 非综合征性唇腭裂是口面部常见的先天性畸形之一,近年来,很多学者对非综合征性唇腭裂患儿及其家属的皮纹学特征进行了一系列研究,并且已经获得了一定的成果。本文就皮纹学与非综合征性唇腭裂等相关遗传疾病、口腔疾病的关系及其流行病学特征等研究进展作一综述。

[关键词] 非综合征性; 唇裂; 腭裂; 皮纹学

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**Research progress on dermatoglyphic features in non-syndromic cleft lip and palate** Ma Hongfang, Shi Jiayu, Shi Bing. (Dept. of Cleft Lip and Palate Surgery, West China Hospital of Stomatology, Sichuan University, Chengdu 610041, China)

[Abstract] Non-syndromic cleft lip with or without cleft palate is a common birth defect. The relationship between cleft lip and palate and dermatoglyphic features is widely recognized, frequently studied, and poorly understood. We summarized a select group of articles on this topic, and discussed the current understanding of dermatoglyphic features in individuals with cleft and their parents, as well as the relative heredity diseases, oral diseases, epidemiology in this article.

[Key words] non-syndromic; cleft lip; cleft palate; dermatoglyphics

皮纹是手指、手掌及脚掌等处皮嵴纹的形态,是由皮肤表皮层分化后所形成的隆起部分,大多是呈长条弯曲状且相互平行的纹线。皮纹形成始于胚胎第7周,第12到第13周时达到形成高峰<sup>[1]</sup>,之后随着人体的发育成熟,皮纹会不断扩大,但其纹型始终不变。皮纹的发育过程受基因和环境的双重影响,在其分化期间,如果遇到特殊的遗传和环境变化,则会产生不同的皮纹模式。唇腭裂也属基因和环境双重因素影响所发生的疾病,故唇腭裂与皮纹的形成具有一定共性。不少国内外遗传学研究结果均提示,人类皮纹异常与唇腭裂的发生存在一定的相关性,应用皮纹学预测唇腭裂的发生可能具有一定的实际意义。

## 1 皮纹学

皮纹在一定程度上可以反映个体出生前的发育情况,并且其纹路终身不变。Cummins<sup>[2]</sup>首先创立了“皮纹学”这一术语,为皮纹学特别是医学

皮纹学奠定了基础。人体的皮纹主要指手纹(包括指纹和掌纹)和足纹,其中手纹的获取较为方便,因此大多以手纹作为研究对象。Cummins将指纹模式分为弓型(分为筒弓和帐弓)、箕型(分为尺箕和桡箕)和斗型(分为简单斗和双斗);而对于掌纹的分析,则多从atd角、ab嵴线数、大小鱼际花纹、指间花纹、掌纹线等方面进行研究。

## 2 皮纹与遗传病的关系

多种先天性疾病均存在皮纹异常,以染色体异常疾病的皮纹学表现最为明显。Matsuyama等<sup>[3]</sup>发现,伴唇腭裂的唐氏综合征患儿及其母亲的皮纹常出现弓型增加、斗型减少的现象,而其父亲的斗型和尺箕则明显减少。Masjkey等<sup>[4]</sup>发现,唐氏综合征患儿皮纹的斗型减少,箕型增加,atd角和adt角会出现明显改变。另有许多文献报道各种肿瘤<sup>[5-7]</sup>、先天性高血压<sup>[8-9]</sup>、先天性心脏病<sup>[10]</sup>、各种综合征<sup>[11-13]</sup>患者均有皮纹上的异常表现。

## 3 皮纹与口腔疾病的关系

某些口腔疾病也存在一些皮纹上的异常表现。

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Madan等<sup>[14]</sup>发现,在无龋儿童中尤其是女性,其皮纹的尺箕出现率最高;在患龋儿童中,斗型皮纹模式最多,尤以右手第三指最为显著,表现为较少的总嵴线数。Tikare等<sup>[15]</sup>发现,1类和2类错颌畸形患者相对于无错颌畸形人群来说,斗型皮纹的出现率有明显差异。

#### 4 流行病学研究

目前关于非综合征性唇腭裂的皮纹学报道较少,研究对象为美国人的有2篇<sup>[16-17]</sup>、日本人的1篇<sup>[18]</sup>、印度人的7篇<sup>[19-24]</sup>、比利时人的1篇<sup>[25]</sup>、澳大利亚人的1篇<sup>[26]</sup>、以色列人的1篇<sup>[27]</sup>、中国人和菲律宾人的3篇<sup>[28-30]</sup>、伊朗人的1篇<sup>[31]</sup>。

Woolf等<sup>[16-17]</sup>分析了美国47名唇腭裂患者后报道,有家族史的唇腭裂患儿及其表型正常的父母和兄弟姐妹的atd角不对称性和ab嵴线数的不对称性,明显高于无家族史表型正常的家庭。

Kanematsu等<sup>[18]</sup>在对唇腭裂组与对照组的比较中发现,皮纹异常和皮纹异常一致性在患儿及其父母中的出现率有所增高;在分析唇腭裂组时还发现,当唇腭裂患儿父母一方有患唇腭裂时,患儿及其患有唇腭裂父母的皮纹异常率均增高,皮纹异常一致性也增高。De Bie等<sup>[25]</sup>对比研究认为,非综合征性唇腭裂患儿与正常儿童间不存在明显差异,但在染色体异常所致的综合征患儿中存在异常的皮纹模式。

有研究<sup>[21-24]</sup>发现,唇裂伴发或不伴发腭裂患者皮纹的尺箕和桡箕明显增多,而指间花纹出现率降低,猿线和悉尼线明显增多;宽atd角( $>30^\circ$ )的出现率增高,皮纹的波动不对称性也增多。

Kobyliansky等<sup>[27]</sup>通过研究得出,唇腭裂患儿的皮纹波动性增加、皮纹的性二态性降低,而其父母的皮纹学特征值与健康对照组类似。Mathew等<sup>[1]</sup>比较了唇腭裂患儿和正常儿童后发现,唇腭裂患儿皮纹的尺箕增多,atd角和atd角的不对称性均变大。Neiswanger等<sup>[28]</sup>研究后发现,中国唇腭裂患儿与其表型正常家属及正常儿童,在各指纹模式的出现率、总嵴线数、atd角、atd角不对称性上均未见明显差异,但有家族史的唇腭裂患儿比无家族史的唇腭裂患儿、表型正常的家属、对照组,在各项指标的不对称性上均有明显的增多表现。Scott等<sup>[29]</sup>研究后发现,菲律宾唇腭裂患儿皮纹的弓型、尺箕出现率明显增多,同时斗型出现率明显减少,尤以女性为甚;唇腭裂患儿与其

表型正常家属相比而言,其皮纹的波动不对称性存在明显差异,并且唇腭裂患儿家属的各种指纹模式出现率介于患儿与健康对照组之间。Scott等<sup>[30]</sup>比较了两地患者后发现,皮纹模式在中国人和菲律宾人中的表现不同,中国人皮纹的桡箕增多;而菲律宾人的弓型和尺箕出现率增高,斗型出现率降低;中国人皮纹的尺箕和斗型在性别上存在明显差异,而菲律宾人仅在斗型上存在明显的性别差异。Jahanbin等<sup>[31]</sup>通过回顾性分析得出,非综合征性唇腭裂患儿父亲皮纹的弓型出现率较高,而其母亲的弓型出现率未见明显改变,双亲的atd角不对称性均有所增高。

#### 5 皮纹与发育及人种的关系

由于特定的遗传和环境因素的影响,发育不稳定可能对发育裂隙的产生及其相关基因产生一定的作用<sup>[16-17]</sup>。发育不稳定可以被理解为机体在发育过程中,对各种遗传或环境干扰因素缓冲能力的下降,在皮纹模式上表现为皮纹模式出现率的变化或高度的波动不对称性。

现研究认为,唇腭裂患儿皮纹的弓型出现率有所增高,斗型的出现率降低。Babler提出,缓慢形成的以弓型为主体的皮纹模式,可能预示了皮纹在形成阶段存在发育受限或发育紊乱的现象。出生前若接触了致畸性或致发育受限的外界刺激,均会增加弓型皮纹的出现率。在唇腭裂患儿及其家属中,以弓型为主的皮纹模式或较为少见的皮纹模式(如放射环状桡箕)存在较高的出现率,这提示了唇腭裂患儿可能存在产前个体发育障碍或发育迟缓等现象。

有文献<sup>[16-17 21-24 27-29 31]</sup>报道,皮纹学的波动不对称性在唇腭裂患儿中的发生率存在明显升高的趋势。

值得一提的是,有学者<sup>[29]</sup>报道,唇腭裂患儿表型正常家属的各指纹模式的出现率介于患儿与健康对照组之间,这可能是由于唇腭裂患儿家族中存在着导致指纹模式异常的等位基因。

现已得出的各项研究报道存在着研究结果相互冲突的问题,这可能是由于唇腭裂患儿及其家属的指纹类型具有人种特异性所致<sup>[30]</sup>。

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