心血管学组

以急性心力衰竭为首发症状的儿童风湿性心脏炎 10 例临床分析

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【摘 要】

目的： 探讨急性心力衰竭为首发症状的儿童风湿性心脏炎的临床表现、病变特点、和治疗预后，以提高临床医师对该病的认识。

方法：回顾性分析 2015 年 1 月～2022 年 2 月在深圳市儿童医院诊治的以急性心力衰竭为首发症状的急性风湿性心脏炎患儿共 10 例，采集临床资料、实验室检查以及影像资料和诊治预后资料。

结果：10 例患儿男性 4 例，女性 6 例，平均就诊年龄 9.1 岁（3 岁 1 月~11 岁 3 月），起病时间 9.3±3.2 天（4~14 天）。10 例患儿中循环系统症状胸痛 4 例，活动耐力降低 5 例；4 例有关节症状，5 例有呼吸道症状，4 例有消化道症状。超声心动图检查示所有患者均有二尖瓣病变，以二尖瓣关闭不全最常见，其中二尖瓣联合病变 4 例，单纯二尖瓣关闭不全 3 例，二尖瓣病变伴主动脉瓣关闭不全 4 例，二尖瓣病变伴三尖瓣关闭不全 1 例。所有患者均进行了抗风湿治疗、抗感染治疗和抗心衰的内科治疗，所有患者经内科治疗后心功能改善，瓣膜病变减轻。随访 1~4 年，6 例遗留慢性风湿性心脏瓣膜病，1 例出现严重二尖瓣关闭不全需外科手术治疗。

结果：以急性心力衰竭为首发症状的风湿性心脏炎，具有临床发病早，除外风湿热典型表现外，常伴有消化道和呼吸道表现；主要病变为二尖瓣病变，超声心动图有助于早期诊断。内科治疗有效，但风湿性心脏瓣膜病后遗症发生率仍较高。

关键词；风湿热；心脏炎；二尖瓣关闭不全; 超声心动图

Construction of A Facial Recognition Assisted-Diagnostic Model for Noonan Syndrome by Merging Virtual Faces ： A novel approach for few-shot learning in medical fields

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Abstract

Background: Genetic syndromes (GSs) are easily misdiagnosed due to their low incidence and complex phenotypes. Most GSs cases such as Noonan Syndrome(NS) have characteristic craniofacial appearances, simultaneously facial dysmorphic features in children with the same genetic syndrome are usually similar. Thus, facial recognition can be used for GSs diagnosis. However, face samples of rare GSs in the real world are difficult to obtain and few-shot learning still remains great challenge in this issue. In recent years, face merging technology has developed in the field of facial recognition. Virtual faces can be generated by merging different facial photograghs. In this paper, virtual faces with special morbid features achieve to be generated by merging dysmorphic faces of NS with normal faces. Consequently, the facial dataset of NS will be augmented.The purpose of this study was to construct a facial recognition assisted-diagnostic model for NS by merging virtual faces to investigate a novel approach for few-shot learning in medical fields.

Methods:A total of 60 frontal facial photos were collected from 60 children with NS in Guangdong Provincial People's Hospital from Jan 2018 to Dec 2021. In the meantime，460 normal facial photos were collected from 460 other children for control group. Only one frontal facial image was selected for each participant. 20 NS facial photos and 20 control facial photos were randomly selected for image merging. Each of the 20 NS photos was merged with 20 different normal facial photos by a merging face software: Face^{++}(Megvii, 2015) , thereby altogether generating 400 virtual faces. The virtual images mixed with control images were shown to the clinicians to assess the image quality. A ResNet-50 framework was pretrained by transfer learning methods, and two NS facial recognition model based on the Resnet-50 was constructed respectively by 400 virtual NS faces and 20 actual NS faces. The performance of the two Resnet-50 models was evaluated by the rest 40 real NS facial photos and 40 normal facial photos. Comparison of ResNet-50 model to six physicians were also performed.

Results: A total of 400 virtual images were generated from 20 facial photographs with NS and 20 control photographs by Face^{++} . 60 virtual faces and 60 real faces were completed the manual recognition. With 96.5% proportion of real faces recognized as actual faces, 93% virtual faces were identified as actual （ P ＞0.05 ） . In the manual recognition for identifying virtual NS faces as NS , the average recognition rate was 33.75% for junior physicians and 70% for senior physicians. There was no statistical difference between virtual NS faces and actual NS faces .

The ResNet-50 model constructed by virtual images achieved the highest accuracy of 0.9375, sensitivity (recall rate) of 0.9000, specificity of 0.9750, precision of 0.9730, F1 of 0.9351, and AUC of 0.9603 （ 0.95CI： 0.9161-1.0000) for NS screening, which was signifcantly higher than that achieved by the controll recognition model based on actual faces and human experts.

Conclusions: This study highlighted the feasibility of generating virtual faces by Face^{++} to amplify the NS facial sample size. The ResNet-50 facial recognition model trained by virtual samples may play a prominent role in other rare GSs screening in further clinical practice.

Keywords: Facial recognition, Noonan Syndrome, Genetic syndrome, Rare diseases, Artifcial intelligence, Deep learning.

婴儿型肝血管内皮细胞瘤并发心力衰竭患儿的临床特点及随访研究

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【摘 要】

目的：总结婴儿型肝血管内皮细胞瘤（IHHE）并发充血性心力衰竭患儿的临床特征、治疗及预后。

方法：对 2012 年 12 月至 2017 年 12 月在广州市妇女儿童医疗中心诊断为 IHHE 并发心力衰竭患儿的临床资料进行回顾性分析，总结患儿一般资料、症状、体征、治疗和随访情况。

结果：本组病例共 15 例，其中男 9 例，女 6 例，年龄 1 天～7 月（中位年龄 36 天），体重 2.6kg～6.7kg（中位体重 4.2kg）。其中 9 例以气促、呼吸困难、哭闹后口唇发绀等表现为首发症状，另 6 例以气促、腹部膨隆为主要症状。2 例合并卡-梅综合征。所有患儿均使用强心、利尿治疗，3 例予波生坦或前列地尔降肺动脉压力治疗。1 例行肝动脉栓塞术联合甲泼尼龙和心得安治疗，11 例予心得安和/或甲泼尼龙治疗。死亡 2 例，其余 13 例患儿经治疗后肝脏肿瘤体积明显缩小，心力衰竭症状明显改善。2 例患儿随访过程中失访，11 例患儿随访 8～65 个月，中位随访 31 个月，所有患儿均无明显症状，复查超声心动图提示心脏大小、心功能、肺动脉压力恢复正常，所需时间为 3 个月（15 天～11 月）。

结论：IHHE 可以心力衰竭为首发症状，临床上不明原因心力衰竭的患儿应积极检查明确是否存在 IHHE，进行早期积极干预可明显改善患儿症状，可联合使用抗心衰治疗和心得安、甲泼尼龙等治疗，如能渡过急性期，中远期预后良好。

关键词：心力衰竭；婴儿型肝血管内皮细胞瘤；治疗；随访

12 月龄以下婴儿川崎病的临床特征：一项单中心十年回顾性研究

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【摘 要】

目的： 目前关于 12 个月以下的川崎病 (KD) 临床表现不典型。本研究旨在分析中国南方婴儿队列中 KD 的临床和实验室特征。

方法：对 2011 年 1 月至 2020 年 12 月间发病时年龄小于 12 月龄的 KD 儿童进行了回顾性分析。临床数据、实验室参数、治疗和结果都收集在川崎病专病数据库中。

结果:在 749 名 KD 患者中，174 名 (23.2%) 小于 12 月龄。6 个月以下患者 67 例（38.5%），3 个月以下患者 29 例（16.7%）。中位年龄为 （6.88+3.05）个月。诊断的平均时间为 （5.4+2.15）天，在无冠状动脉损伤组及完全性川崎病组中更长，分别是（6.88+ 3.57 ）天和（6.12+2.06）天。120 名患者 (69%) 存在结膜充血；133 人 (76.4%) 出现皮疹；肢体变化 130（74.7%）；161 例（92.5%）粘膜改变，仅 150 例（86.2%）淋巴结肿大。粘膜变化是不完全形式中最不常见的特征（86.8%）。76 名患者 (43.6%) 患有不完全 KD。不完全 KD 确诊时发热小于 5 天有 26 人（34.2%），较完全 KD 明显增多，5 项典型症状中皮疹最常见。26 人 (14.9%) 有心脏受累，其中 13 人 (50%) 有不完全形式，3 个月以下患者比例最高（20.68%）。CAL- KD 患者中 PLT 较高，白蛋白较低。所有患者均接受 IVIG 治疗，17 名（9.7%）患者无反应。

结论:在我们的队列中，更早发现冠状动脉损伤和不完全 KD。发现诊断不完全 KD 比例较高，其诊断发热时间多小于 5 天，皮疹是最常见的症状。冠状动脉损伤发生率是 14.9%，其中 3 个月以下患者最容易发生冠状动脉损伤。因此，我们认为婴儿 KD 并不少见，特别是 3 个月以下婴儿发热伴皮疹，需注意不完全 KD。

关键词：川崎病，婴儿，冠状动损伤，不完全 KD