966－977.
[2] 袁犁，周琦，徐发良，等. 卵巢癌紫杉醇耐药细胞耐药性和髓样分化因子88关系的研究[J]. 重庆医科大学学报，2012，37(7)：570－574.
[3] Zhande R, Dauphinee S M, Thomas J A, *et al*. FADD negatively regulates lipopolysaccharide signaling by impairing interleukin-1 receptor-associated kinase 1-MyD88 interaction[J]. Mol Cell Biol, 2007, 27(21): 7394－7404.
[4] Gelman A E, LaRosa D F, Zhang J, *et al*. The adaptor molecule MyD88 activates PI-3 kinase signaling in CD4$^+$ T cells and enables CpG oligodeoxynucleotide-mediated costimulation[J]. Immunity, 2006, 25(5): 783－793.
[5] Davis C N, Mann E, Behrens M M, *et al*. MyD88-dependent and -independent signaling by IL-1 in neurons probed by bifunctional Toll/IL-1 receptor domain/BB-loop mimetics[J]. Proc Natl Acad Sci U S A, 2006, 103(8): 2953－2958.
[6] Kelly M G, Alvero A B, Chen R, *et al*. TLR-4 signaling promotes tumor growth and paclitaxel chemoresistance in ovarian cancer[J]. Cancer Res, 2006, 66(7): 3859－3868.
[7] Mao H L, Liu P S, Zheng J F, *et al*. Transfection of Smac/DIABLO sensitizes drug-resistant tumor cells to TRAIL or paclitaxel-induced apoptosis *in vitro*[J]. Pharmacol Res, 2007, 56(6): 483－492.
[8] Zhu Y, Huang J M, Zhang G N, *et al*. Prognostic significance of MyD88 expression by human epithelial ovarian carcinoma cells[J]. J Transl Med, 2012, 10: 77.
[9] Chen H, Jiang Z. The essential adaptors of innate immune signaling[J]. Protein Cell, 2013, 4(1): 27－39.
[10] Chun J, Choi R J, Khan S, *et al*. Alantolactone suppresses inducible nitric oxide synthase and cyclooxygenase－2 expression by down-regulating NF-kappaB, MAPK and AP-1 *via* the MyD88 signaling pathway in LPS-activated RAW 264.7 cells[J]. Int Immunopharmacol, 2012, 14(4): 375－383.
[11] Bauerfeld C P, Rastogi R, Pirockinaite G, *et al*. TLR4-mediated AKT activation is MyD88/TRIF dependent and critical for induction of oxidative phosphorylation and mitochondrial transcription factor A in murine macrophages[J]. J Immunol, 2012, 188(6): 2847－2857.
[12] Zhang X, Pino G M, Shephard F, *et al*. Distinct control of MyD88 adapter-dependent and Akt kinase-regulated responses by the interleukin (IL)-1RI co-receptor, TILRR[J]. J Biol Chem, 2012, 287(15): 12348－12352.
[13] Bosmann M, Patel V R, Russkamp N F, *et al*. MyD88-dependent production of IL-17F is modulated by the anaphylatoxin C5a *via* the Akt signaling pathway[J]. FASEB J, 2011, 25(12): 4222－4232.
[14] Foster J G, Blunt M D, Carter E, *et al*. Inhibition of PI3K signaling spurs new therapeutic opportunities in inflammatory/autoimmune diseases and hematological malignancies[J]. Pharmacol Rev, 2012, 64(4): 1027－1054.
[15] Blajecka K, Borgstrom A, Arcaro A. Phosphatidylinositol 3-kinase isoforms as novel drug targets[J]. Curr Drug Targets, 2011, 12(7): 1056－1081.
[16] Ieiri I. Functional significance of genetic polymorphisms in P-glycoprotein (MDR1, ABCB1) and breast cancer resistance protein (BCRP, ABCG2)[J]. Drug Metab Pharmacokinet, 2012, 27(1): 85－105.

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个案与短篇

舞蹈病-棘状红细胞增多症1例

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[**关键词**]　舞蹈病-棘状红细胞增多症；诊断；鉴别诊断
[**中图法分类号**]　R742.2　[**文献标志码**]　B

舞蹈病-棘状红细胞增多症(chorea-acanthocytosis，ChAc)是一种罕见的常染色体隐性遗传病。常伴有棘红细胞增多，主要临床表现为突发的、不规则的、类似亨廷顿舞蹈病样的不自主运动以及特征性的口面部不自主运动，常导致牙齿磨损、舌咬伤、发出怪声、构音障碍等。本科诊治1例，现报告如下。

1　临床资料

患者，女性，36岁，因“言语不清、行走不稳3年余，加重半年”于2012年10月7日入院。患者于3年前无明显诱因出现言语不清，行走不稳，伴不自主磨牙、不自主右肩耸动，余未诉特殊不适；不影响日常生活，未就诊。近半年患者感言语不清、行走不稳较前进一步加重，行走时容易跌倒，仍伴不自主磨牙、耸右肩，影响日常生活，无明显肢体舞蹈样动作，无咬舌和咬指头、发出怪声、吞咽困难等情况，进食时无明显诱发伸舌和躯干部伸屈运动。既往史、个人史及家族史无特殊。

查体：患者上下牙冠变平，意识清楚，查体合作，言语含糊，对答切题，四肢肌肉无萎缩，四肢肌力正常，四肢肌张力下降，可见不自主磨牙，不自主耸动右肩，指鼻试验、闭目难立征阴性，四肢腱反射减弱，双侧病理征阴性。辅助检查：颅脑MRI示：双侧侧脑室前角稍增宽，尾状核萎缩。行焦虑自评量表(SAS)及抑郁自评量表(SDS)示：重度焦虑、抑郁。脑电图示：成人正常脑电图。血常规、大便常规、肾功能、血沉、甲状腺功能、铜蓝蛋白正常。乙肝三对未见明显异常。抗O试验及类风湿因子阴性。肝功能示：ALT 104 IU/L，AST 125 IU/L。外周血涂片可见棘红细胞(>3%)。诊断：舞蹈病-棘状红细胞增多症。治疗：入院后给予口服苯妥英钠片0.1 g口服，3次/d，枸橼酸坦度罗酮10 mg口服，3次/d抗焦虑抑郁治疗。患者不自主运动较入院明显减少出院。

2　讨论

舞蹈病-棘红细胞增多症是一种罕见的神经系统变性疾病，可累及多个系统。本病发病年龄8～62岁，平均35岁。临

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参考文献：

[1] Osaka M, Rowley J D, Zeleznik-Le N J. MSF (MLL septin-like fusion), a fusion partner gene of MLL, in a therapy-related acute myeloid leukemia with a t(11;17)(q23;q25)[J]. Proc Natl Acad Sci U S A, 1999, 96(11): 6428-6433.

[2] Chan D C, Bedford M T, Leder P. Formin binding proteins bear WWP/WW domains that bind proline-rich peptides and functionally resemble SH3 domains[J]. EMBO J, 1996, 15(5): 1045-1054.

[3] Megonigal M D, Rappaport E F, Jones D H, *et al*. t(11;22)(q23;q11.2) In acute myeloid leukemia of infant twins fuses MLL with hCDCrel, a cell division cycle gene in the genomic region of deletion in DiGeorge and velocardiofacial syndromes[J]. Proc Natl Acad Sci U S A, 1998, 95(11): 6413-6418.

[4] Fuchs U, Rehkamp G, Haas O A, *et al*. The human formin-binding protein 17 (FBP17) interacts with sorting nexin, SNX2, and is an MLL-fusion partner in acute myelogeneous leukemia[J]. Proc Natl Acad Sci U S A, 2001, 98(15): 8756-8761.

[5] Joh T, Yamamoto K, Kagami Y, *et al*. Chimeric MLL products with a Ras binding cytoplasmic protein AF6 involved in t(6;11) (q27;q23) leukemia localize in the nucleus[J]. Oncogene, 1997, 15(14): 1681-1687.

[6] Kamioka Y, Fukuhara S, Sawa H, *et al*. A novel dynamin-associating molecule, formin-binding protein 17, induces tubular membrane invaginations and participates in endocytosis[J]. J Biol Chem, 2004, 279(38): 40091-40099.

[7] Takano K, Toyooka K, Suetsugu S. EFC/F-BAR proteins and the N-WASP-WIP complex induce membrane curvature-dependent actin polymerization[J]. EMBO J, 2008, 27(21): 2817-2828.

[8] Tsuboi S, Takada H, Hara T, *et al*. FBP17 Mediates a Common Molecular Step in the Formation of Podosomes and Phagocytic Cups in Macrophages[J]. J Biol Chem, 2009, 284(13): 8545-8556.

[9] Tsujita K, Kondo A, Kurisu S, *et al*. Antagonistic regulation of F-BAR protein assemblies controls actin polymerization during podosome formation[J]. J Cell Sci, 2013, 126(Pt 10): 2267-2278.

[10] 林明明，张乾英，王蕴红，等. 沉默FNBP1表达诱导7703细胞形态重塑[J]. 激光生物学报，2010，19(5)：669-672.

[11] Hynes R O. Integrins: bidirectional, allosteric signaling machines[J]. Cell, 2002, 110(6): 673-687.

[12] Shimaoka M, Takagi J, Springer T A. Conformtional regulation of integrin structure and function[J]. Annu Rev Biophys Biomol Struct, 2002, 31: 485-516.

[13] Firat-Karalar E N, Welch M D. New mechanisms and functions of actin nucleation[J]. Curr Opin Cell Biol, 2011, 23(1): 4-13.

[14] Tsujita K, Suetsugu S, Sasaki N, *et al*. Coordination between the actin cytoskeleton and membrane deformation by a novel membrane tubulation domain of PCH proteins is involved in endocytosis[J]. J Cell Biol, 2006, 172(2): 269-279.

[15] Tskvitaria-Fuller I, Rozelle A L, Yin H L, *et al*. Regulation of sustained actin dynamics by the TCR and costimulation as a mechanism of receptor localization[J]. J Immunol, 2003, 171(5): 2287-2295.

[16] 曹文枫，孙保存. Rho蛋白与细胞骨架调节的细胞运动[J]. 中国肿瘤临床，2009，36(14)：835-837.

[17] 邵静，王红兵，杨力，等. Rho蛋白对肿瘤细胞骨架活动及生长调节的影响[J]. 生物医学工程学杂志，2008，25(6)：1462-1465.

[18] Narumiya S, Ishizaki T, Watanabe N. Rho effectors and reorganization of actin cytoskeleton[J]. FEBS Lett, 1997, 410(1): 68-72.

[19] 吴平平，苏昀，金治，等. Rho A蛋白通路在DLC-1基因调控人结肠癌HT29细胞周期中的作用及其机制[J]. 东南大学学报：医学版，2009，28(4)：247-251.

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床表现为四肢突发的、不规则的类亨廷顿舞蹈病样动作，不自主运动常累及口面部，导致咬舌和咬指头、发出怪声、吞咽困难和构音障碍等。尤其食物诱发伸舌和躯干部伸屈运动几乎不见于其他舞蹈症[1]。肌张力低下亦很常见。少数病例也可表现为帕金森综合征[2]。以认知功能障碍、精神症状以及智力减退等大脑皮质受损为主要临床表现者也较常见[3]。实验室检查主要包括外周血涂片可见棘红细胞，一般棘红细胞比例在3%以上才有意义，部分患者仅在疾病晚期出现棘红细胞[4]，故未见棘红细胞不能除外此病。影像学表现主要是CT或MRI示侧脑室前角增宽，尾状核和豆状核萎缩，该患者颅脑MRI示：侧脑室前角稍增宽，尾状核萎缩。这与文献[5]报道相同。根据典型临床表现和实验室检查发现棘红细胞一般可诊断。鉴别诊断：①无β脂蛋白血症是一种常染色体隐性遗传性病，因脂肪代谢障碍导致缺乏维生素E，临床主要表现为脊髓小脑变性、周围神经病变及色素性视网膜炎。②泛酸激酶相关性神经变性也是一种常染色体隐性遗传病。一般10岁以后起病，因铁代谢障碍，基底节区铁沉积，从而出现构音障碍，色素性视网膜病。其颅脑MRI显示特征性的“虎眼征”（苍白球区域低信号影伴中心高信号影）。③亨廷顿舞蹈病则是一种常染色体显性遗传病，常有家族史，无癫痫发作，无周围神经及肌病表现，肢体舞蹈幅度大，无食物诱发伸舌，周围血红细胞一般无棘红细胞发现。

该病目前缺乏有效治疗方法，主要针对舞蹈症状，采用药物或者脑深部电刺激等方法予以对症治疗，并通过心理治疗、功能锻炼及康复训练等进一步改善患者的症状。

参考文献：

[1] Bader B, Walker R H, Vogel M, *et al*. Tongue protrusion and feeding dystonia: a hallmark of chorea-acanthocytosis[J]. Mov Disord, 2010, 25(1): 127-129.

[2] Bostantjopoulou S, Katsarou Z, Kazis A, *et al*. Neuroacanthocytosis presenting as parkinsonism[J]. Mov Disord, 2000, 15(6): 1271-1273.

[3] Danek A, Sheesley L, Tierney M, *et al*. Cognitive findings in patients with chorea-acanthocytosis[J]. J Int Neuropsychol Soc, 2003, 9: 528.

[4] Scheid R, Bader B, Ott D V, *et al*. Development of mesial temporal lobe epilepsy in chorea-acanthocytosis[J]. Neurology, 2009, 73(17): 1419-1422.

[5] 周祥琴，关鸿志，史向松，等. 神经棘红细胞增多症的临床及神经影像学特征[J]. 中华神经科杂志，2012，45(2)：112-115.

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