

Neuroacanthocytosis in China: A Review of Published Reports

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Abstract

Background: Neuroacanthocytosis (NA) syndromes are a group of rare diseases characterized by the presence of acanthocytes and neuronal multisystem pathology, including chorea-acanthocytosis (ChAc), McLeod syndrome (MLS), Huntington's disease-like 2 (HDL-2), and pantothenate kinase-associated neurodegeneration (PKAN). China has the largest population in the world, which makes it a good location for investigating rare diseases like NA.

Methods: We searched Medline, ISI Proceedings, China National Knowledge Infrastructure, and Wanfang Data for literature published through December 31, 2013 for all the published Chinese NA case reports and extracted the clinical and laboratory findings.

Results: A total of 42 studies describing 66 cases were found to be eligible for inclusion. Age of symptom onset ranged from 5 to 74 years. The most common findings included hyperkinetic movements (88%), orofacial dyskinesia (80%), dystonia (67%), and dysarthria (68%), as well as caudate atrophy or enlarged lateral ventricles on neuroimaging (64%), and elevated creatine kinase (52%). Most cases were not confirmed by any specific molecular tests. Only two cases were genetically studied and diagnosed as ChAc or MLS.

Discussion: In view of the prevalence of NA syndromes in other countries, the number of patients in China appears to be underestimated. Chinese NA patients may benefit from the establishment of networks that offer specific diagnoses and care for rare diseases.

Keywords: Neuroacanthocytosis, chorea-acanthocytosis, McLeod syndrome, neurodegeneration with brain iron accumulation, China

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Introduction

Neuroacanthocytosis (NA), an umbrella term for a group of rare diseases characterized by misshapen erythrocytes (acanthocytes) and neuronal multisystem pathology, includes diseases such as autosomal recessive chorea-acanthocytosis (ChAc), X-linked McLeod syndrome (MLS), Huntington's disease-like 2 (HDL-2), and pantothenate kinase-associated neurodegeneration (PKAN).¹ NA was first described as Levine-Critchley syndrome on the basis of reports by Levine and Critchley in two families from North America in the 1960s.^{2,3} Although the clinical manifestations of NA resemble Huntington's disease (HD), there are clear differences in the mode of inheritance and neuropathological changes. In the last decade, molecular and protein methods have rapidly developed for the diagnosis of different NA subtypes.

China has the largest population in the world (1.35 billion), making it a suitable location for investigating rare diseases like NA. The first Chinese NA literature is a translation of the paper by Sakai et al in 1981.^{4,5} Over the last 30 years, few Chinese NA case reports have been published.^{6–53} Moreover, communication with foreign scholars was limited because most of the reports were published in Chinese. ChAc has been reported in many different ethnic groups, and the relatively high prevalence in Japan appears to be due to a founder mutation.⁵⁴ In light of the similar East Asian backgrounds in the two countries, we suspect that in China NA syndromes are underdiagnosed, leading to a significant underestimation of the prevalence.

In the current study, we systematically reviewed Chinese NA epidemiology by searching for published NA cases. We aimed to

Table 1. Baseline Information on NA Cases from China

Ref.	Place of Diagnosis	Year of Publication	No. Cases	Sex	AOO	Initial Symptoms	Acanthocytes (%)	Reported Diagnosis	Our Diagnostic Hypothesis
6	Anhui	1984	2	2M	19, 27	Orof. (1), MD/Orof. (1)	23%, 3%	ChAc	
7	Anhui	1987	2	2M	26, 32	DYA (1), MD/Orof. (1)	10%	ChAc	
8	Anhui	2000	2	2F	45, 46	MD (1), PSY (1)	35%, 17%	ChAc	
9	Anhui	2004	1	1M	39	Orof./MD	4–7%	ChAc	
10	Beijing	2005	1	1F	n.a.	MD	4%	ChAc	
11–14	Beijing	2005, 2012	8	2M, 6F	10–35	Orof. (4), MD (3), PN (1)	>10%	NA	
15	Beijing	2007	2	2M	27, 37	MD (1), EPI (1)	15–25%	ChAc	
16	Beijing	2010	1	1M	11	n.a.	30%	NA	
17, 18	Beijing	2013	1	1M	42	MD/Orof.	>40%	NA	NBIA
19	Beijing	2013	3	3M	16–43	MD, Orof. (2), BP (1)	>30%	NA	
20	Chongqing	2013	1	1F	33	DYA/WI/MD/Orof.	>3%	ChAc	
21	Gansu	2009	1	1M	20	MD	n.a.	NA	
22	Guangdong	2003	1	1M	42	Orof.	30%	ChAc	
23	Helongjiang	1989	1	1F	36	DYA/Orof.	15–20%	ChAc	
24	Helongjiang	1989	2	1M, 1F	9, 11	Orof.	56%, 28%	ChAc	NBIA
25	Helongjiang	1990	1	1F	38	Orof.	40–50%	ChAc	
26, 27	Helongjiang	2003, 2007	1	1F	35	Orof.	28.5%	ChAc	
28	He'nan	2005	1	1F	30	Orof./EPI	25%	NA	
29	He'nan	2011	2	1M, 1F	30, 31	MD (1), Orof. (1)	35%, 25%	ChAc	
30	He'nan	2012	1	1M	43	MD	11%–15%	ChAc	
31	Hong Kong	2013	1	1M	47	GD/MD	16%	MLS*	
32	Hubei	2007	1	1F	28	Orof.	6–8%	NA	
33	Hubei	2013	1	1M	37	n.a.	n.a.	NA	
34	Hu'nan	2013	1	1F	24	n.a.	n.a.	ChAc*	
35	Jiangsu	2012	1	1F	20	MD/Orof.	7.8%	ChAc	
36	Liaoning	1989	1	1M	28	Orof.	84%	ChAc	

Table 1. Continued

Ref.	Place of Diagnosis	Year of Publication	No. Cases	Sex	AOO	Initial Symptoms	Acanthocytes (%)	Reported Diagnosis	Our Diagnostic Hypothesis
37	Neimenggu	2005	1	1M	32	EPI	10%	ChAc	
38	Neimenggu	2007	1	1M	34	MD	10%	ChAc	
39	Neimenggu	2012	1	1M	38	Orof.	5%	ChAc	
40	Qinghai	2006	1	1F	24	MD	18%	NA	
41	Shandong	2001	3	3F	20–21	MD	12%	NA	
42	Shandong	2013	1	1M	32	MD/Orof.	10%	ChAc	
43	Shandong	2013	3	2M, 1F	26–50	MD/Orof. (1), PSY (2)	>30%	ChAc	
44	Shanghai	2008	1	1F	33	MD/Orof.	7–8%	ChAc	
45	Shanghai	2008	1	1M	5	DYT	>30%	NA	NBIA
46	Shanghai	2010	1	1F	55	WI	>20%	NA	
47	Shanghai	2012	3	2M, 1F	31–74	MD (1), GD (2)	10–28%	NA	
48	Shanghai	2013	1	1M	46	WI, DYA	44.9%	NA	
49, 50	Shanxi	2004	3	2M, 1F	26–30	EPI (2), Orof. (1)	12–30%	ChAc	
51	Sichuan	2012	2	2M	17, 18	Orof. (2)	6%, 6%	ChAc	
52	Sichuan	1988	1	1M	25	Orof.	4%	ChAc	
53	Taiwan	2006	1	1F	31	MD	49%	NA	

Abbreviations: *, Gene Confirmed; AOO, Age of Onset; BP, Blepharospasm; ChAc, Chorea-Acanthocytosis; DYA, Dysarthria; DYT, Dystonia; EPI, Epilepsy; F, Female; GD, Gait Disturbance; M, Male; MD, Movement Disorders (Chorea or Hyperkinetic Movements); MLS, McLeod Syndrome; n.a., Not Available; NA, Neuroacanthocytosis; NBIA, Neurodegeneration with Brain Iron Accumulation (Type I/Type II); No., Number; Orof., Orofacial Dyskinesia; PN, Parkinsonism; PSY, Psychiatric Symptoms; Ref., Reference; WI, Walking Instability.

summarize the features of Chinese NA cases in comparison with other countries and identify potential reasons for underestimation.

Methods

We searched Medline (from January 1, 1949 to December 31, 2013), ISI Proceedings (January 1, 1990 to December 31, 2013), China National Knowledge Infrastructure (<http://www.cnki.net>, from January 1, 1979 to December 31, 2013) and Wanfang Data (<http://www.wanfangdata.com.cn>, from January 1, 1984 to December 31, 2013). The search terms in English and their Chinese equivalents included “neuroacanthocytosis,” “chorea-acanthocytosis,” “McLeod syndrome,” “choreoacanthocytosis,” “hereditary acanthocytosis syndrome,” and “Levine-Critchley syndrome.” We also tried to identify cases from cross-references between papers. We only included those

case reports where an explicit diagnosis of NA had been made by the original authors. For inclusion, patients had to be of Chinese origin and diagnosed in China. We extracted information on geographical origin, sex, age of onset, and clinical and laboratory findings from each case.

Results

We identified a total of 48 publications with 66 cases reported (37 males and 29 females) (Table 1). All of these patients had been diagnosed with NA, ChAc, or MLS by the reporting physicians. Only one ChAc case was confirmed by a genetic test (*VPS13A* gene), and no cases were confirmed by Western blot for the protein chorein that is affected in ChAc.⁵⁵ An elevated proportion of acanthocytes in blood samples was regarded as the most important diagnostic clue by Chinese physicians. However, heterogeneous methods for detecting

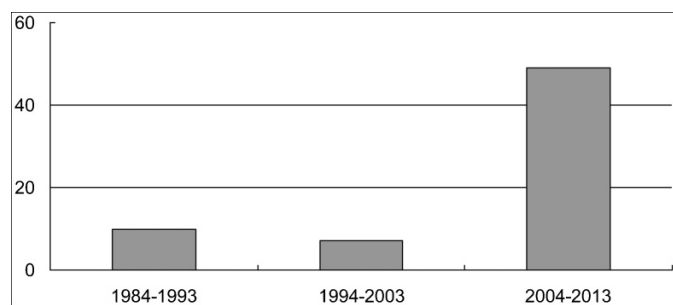


Figure 1. The Number of Chinese NA Cases Reported in the Last Three Decades. In the most recent decade (2004 to 2013) the number of reported cases increased dramatically, being about five times the number documented from 1984 to 1993, and seven times the number

acanthocytosis were used in the different reports, thus affecting the accuracy of the test. Only one MLS case, from Hong Kong, was reported with a corresponding *XK* gene mutation.

In the most recent decade (2004 to 2013) the number of reported cases increased dramatically, being about five times the number documented from 1984 to 1993, and seven times the number documented between 1994 and 2003 (Figure 1). No video recordings were published with the publications reviewed in this study. With respect to the geographic distribution, all cases originated from 19 Chinese provinces, with 16 cases reported in Beijing, 7 cases from Anhui, and 7 cases from Shangdong as the top 3 provinces. The ages of onset ranged from 5 to 74 years and were concentrated in the third and fourth decades ($n = 37, 56\%$) (Figure 2). Common initial symptoms were orofacial dyskinesia ($n = 27, 41\%$) and hyperkinetic limb or trunk movements ($n = 27, 41\%$). Dysarthria, epilepsy, gait disturbance, parkinsonism, and psychiatric symptoms were also reported as the initial symptoms. Diagnoses were mainly made based on the presence of a hyperkinetic movement disorder, elevated acanthocytes in the blood, the pattern of inheritance (recessive), and the exclusion of other possible diseases, such as HD.

Figure 3 summarizes the clinical and laboratory findings reported in Chinese NA cases. The most frequent findings were hyperkinetic movements ($n = 58, 88\%$). Orofacial dyskinesia was reported in 53 patients (80%). In addition, dystonia ($n = 44, 67\%$) and dysarthria (n

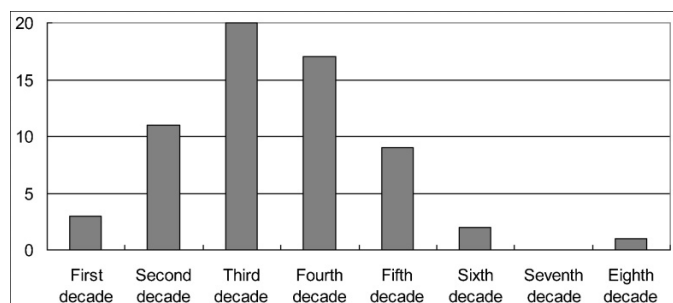


Figure 2. The Distribution of Ages of Onset in Chinese NA Cases. Among the 63 cases with clearly documented ages of onset, most were concentrated in the third and fourth decades (ages 21 to 40).

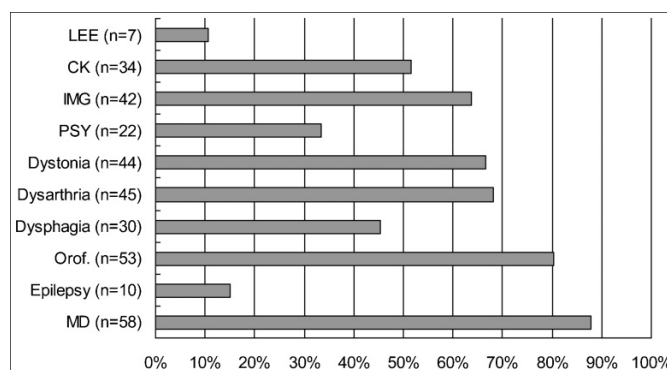


Figure 3. Clinical and Laboratory Findings in Chinese NA Patients (n = 66). Abbreviations: CK, Creatine Kinase Elevation; IMG, Caudate Atrophy or Enlarged Lateral Ventricles on Neuroimaging; LEE, Liver Enzyme Elevation; MD, Movement Disorders (Chorea or Hyperkinetic Movements); NA, Neuroacanthocytosis; Orof., Orofacial Dyskinesia; PSY, Psychiatric Symptoms.

= 45, 68%) were also common. 10 patients (15%) suffered from epilepsy, and 22 patients (33%) exhibited psychiatric symptoms.

With respect to clinical investigations, caudate atrophy or enlarged lateral ventricles on neuroimaging ($n = 42, 64\%$) and elevated CK ($n = 34, 52\%$) were the most frequently reported.

Discussion

Hyperkinetic movements and orofacial dyskinesia were the most common symptoms in this series of Chinese NA cases, reported in 88% and 80% of patients, respectively. This is consistent with the literature in that orofacial dyskinesia appears to be a relatively specific NA symptom, characterized by tongue protrusion and feeding dystonia. Chorea was typical in the early stages and tended to progress to parkinsonism. Parkinsonism has occasionally been reported to be the initial symptom.^{56,57}

We found lower proportions of patients with epilepsy (15%) and psychiatric symptoms (33%) than reported in the literature (50% and 66%, respectively).^{58,59}

In our study, 52% of Chinese NA cases had elevated CK levels, while only 11% of cases exhibited liver enzyme elevation, suggesting that CK is probably a more useful biomarker for NA than acanthocytes or liver enzymes, as is consistent with the literature. However, values within normal ranges are likely to have been omitted in many publications, thus, we could not accurately calculate the proportion of patients with normal CK or liver enzyme levels.⁵⁸

The number of cases reported for 1994–2003 is relatively low in comparison to the numbers for 1984–1993 and 2004–2013. It is possible that some cases were not reported because they were not recognized or were not thought to be of interest after awareness was originally raised regarding the syndrome. It is important to determine whether these low rates of reported cases are due to publication bias and incomplete documentation, or whether these discrepancies are due to ethnic differences.

The clinical diagnosis of NA in China is mainly based on clinical manifestations and an elevated proportion of acanthocytes in the

blood, as well as the exclusion of other possible diseases. Previously this was the standard diagnostic evaluation worldwide (e.g., a Chinese patient diagnosed with ChAc in Singapore in the 1980s).⁶⁰ Recently, however, there have been clear NA cases without acanthocyte elevation.⁶¹

The other forms of NA, such as HDL-2 and PKAN, have not yet been reported in China. However, as some of the NA cases reported had an early age of onset (Table 1), it is likely that PKAN was the correct diagnosis.

With regard to MLS, only one Chinese case has been identified, indicating that it is probably underdiagnosed. Without specific molecular testing, an accurate diagnosis for presumed NA cases is impossible. As an alternative to DNA analyses, chorein detection by Western blot is an inexpensive method for diagnosing ChAc, and Kell antigen phenotyping can be performed to diagnose MLS.^{55,62}

In view of the prevalence of NA syndromes in countries such as the United Kingdom and Germany (approximately 3 in 10 million), the number of NA patients in China should be at least 400 and appears to be underestimated. In addition to raising awareness among Chinese neurologists, the absence of specific molecular tests is the main diagnostic flaw that needs to be addressed. A Chinese network of NA collaboration may improve the diagnosis and treatment for patients affected by this group of incapacitating hyperkinetic movements.

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Note:

Since the acceptance of this article, additional NA cases from China were reported:

Man BL, Yuen YP, Fu YP. The first report of a Chinese family with McLeod syndrome. *BMJ case reports*. 2014. doi:10.1136/bcr-2013-202785

Long X, Xiong N, Guo K, et al. Functional neuroimaging might enable the early diagnosis of neuroacanthocytosis. *Can J Neurol Sci*. 2014;41:402-404