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RESEARCH ARTICLE

Improving the Diagnosis of Underrecognized, Rare Diseases in Asian Populations: Systematic Analysis of Rare Disease Databases

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Rare diseases affect approximately 30 million Americans, but fewer than one in 10 of these patients receive an accurate diagnosis and timely appropriate treatment. Many of these medical conditions disproportionately impact patients of Asian descent as well as other racial and ethnic minorities; however, at the time of this writing, they are not well recognized or sufficiently represented in current medical training curricula or existing scientific literature. To better assess these disparities, we conducted a systematic analysis of the National Organization for Rare Disorders (NORD) and Genetic and Rare Diseases (GARD) Information Center databases to identify rare diseases that are often misdiagnosed in individuals of Asian descent, as well as scientific studies through the PubMed search engine that discuss racial and ethnic disparities in rare clinical diagnoses. Searches in the NORD and GARD databases yielded 52 medical conditions with reported disproportionate prevalence across Asian populations. A subsequent PubMed search regarding these 52 medical conditions identified 133 articles relevant to the potential misdiagnosis and underrecognition of these rare diseases. Overall, there is a paucity of literature on rare diseases, and our findings highlight the need for more research on underrecognized rare diseases that disproportionately impact Asian populations. Future educational programs for medical trainees and practitioners should increase focus on rare diseases in racial and ethnic minority groups to improve diagnosis and minimize disparities in health outcomes.

Key Words: Rare Diseases ▪ Asian Health ▪ Diagnosis ▪ Misdiagnosed Conditions ▪ Health Disparities ▪ Equity

Rare diseases are medical conditions that affect fewer than one in 200,000 people in the U.S (1). Among the approximately 30 million Americans affected by rare diseases, less than one in 10 receive a correct diagnosis and necessary medical treatment (2). Many of these conditions impact Asians, who comprise approximately 7% of the total U.S. population (3), and are projected to become the largest racial and ethnic minority group in the country by mid-century (4, 5). However, across clinical and self-reported metrics, the health status of Asian Americans lags behind their Non-Hispanic White (white) counterparts (6) for numerous reasons. Historically, Western medical literature and education have not focused on rare diseases, especially those that may disproportionately affect people of non-Western descent. Race and ethnicity are social constructs, and those identifying as 'Asian' encompass a large and heterogenous group of diverse backgrounds and cultural identities. These individuals may be at risk for certain conditions due to a complex interplay of biological, genetic, demographic, geographic, and sociocultural factors. With respect to Asians emigrating to the U.S., disease susceptibility can be further nuanced by prenatal and pediatric care, infectious exposures, vaccination history, and adult healthcare received prior to immigration. Patients may also be unaware of previously contracted medical conditions that can persist for years without presenting clinical symptoms. Failure to account for the complexity of these circumstances can lead to incorrect or delayed diagnoses and, therefore,

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POPULAR SCIENTIFIC SUMMARY

- Systematic search of rare disease databases identified 52 medical conditions that can occur at higher rates in Asian populations.
- Some of these rare diseases are not so rare in Asian countries.
- These rare diseases can be underrecognized and misdiagnosed in the United States and other Western countries.

inappropriate or lack of treatment with the potential for patient harm (7–9).

Minority health status is often compromised by institutionalized inequities in employment, income, and education (6, 10–14), as well as the perseverance of cultural norms that discourage healthcare utilization (10, 12–17). Nonetheless, the prevalent 'model minority' myth attributed to Asian Americans masks the distinct challenges of Asians, resulting in the under-emphasis and misreporting of Asian health problems in Western medical literature (11, 13). This knowledge gap can manifest in medical training curricula, and ultimately healthcare provider behavior when treating minority patients (6, 12, 13, 15). Non-Asian health data are often inappropriately applied to Asians, resulting in erroneous generalizations with dire health consequences.

The overall goal of this study is to improve the recognition of rare and potentially neglected conditions in the U.S., which may disproportionately impact patients of Asian descent. While some medical conditions may commonly present in certain areas of Asia, they may be underrecognized, underdiagnosed, or misdiagnosed in the U.S., where they are infrequently seen by clinicians. At present, these racial and ethnic disparities in clinical diagnoses remain largely unexplored. To our knowledge, no study has systematically indexed medical conditions that are commonly misdiagnosed in Asians in the U.S. Therefore, the objectives of this study are to: (1) identify rare diseases in the U.S. that disproportionately affect Asian populations and (2) review published literature that relates to the concept of misdiagnosis of these disproportionately represented rare diseases. The results of this study along with the review of current research raise awareness for these

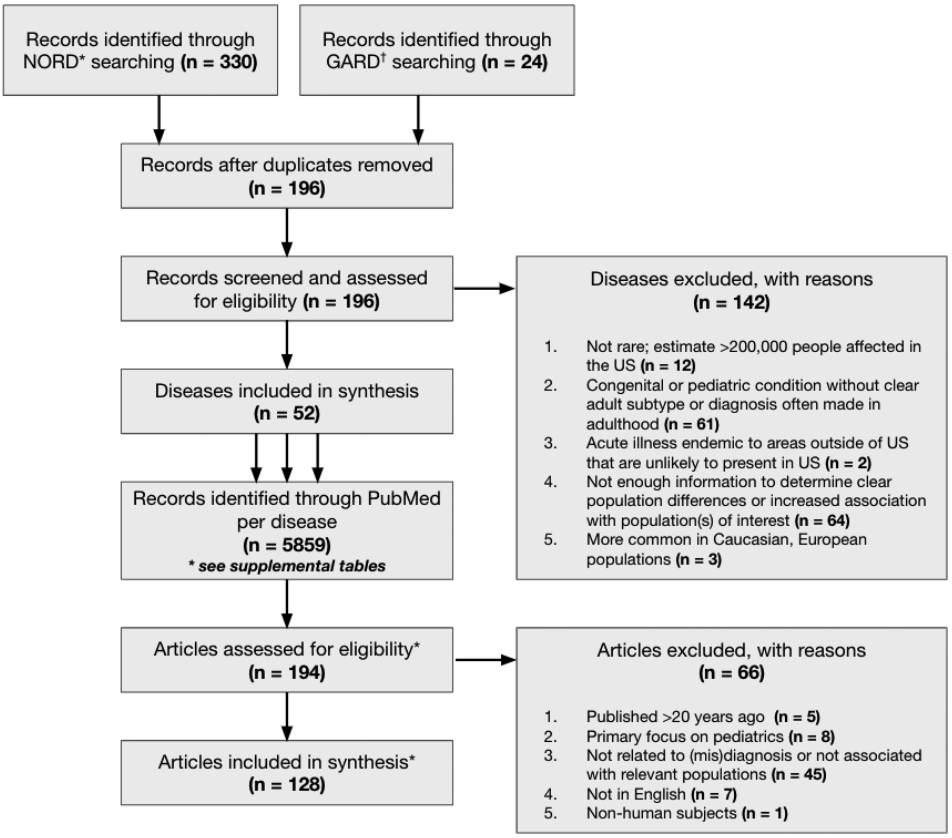

Figure 1. PRISMA-ScR flowchart.

*National Organization for Rare Disorders (NORD).

†Genetic and Rare Diseases (GARD).

conditions and can help clinicians improve differential diagnoses to optimize care for diverse patient populations (8).

METHODS

Information sources and search strategy

The systematic analysis approach undertaken in this study follows the framework of PRISMA-ScR (18) (Preferred Reporting Items for Systematic Reviews and Meta-Analyses extension for Scoping Reviews) (Fig. 1). We first examined disease reports in NORD (National Organization for Rare Diseases) (19) and its related database, GARD (Genetic and Rare Diseases Information Center) (2), for rare diseases that affect fewer than 200,000 people (1). From June to October 2021, authors KA, GM, and LG conducted initial searches in NORD and GARD using a bank of terms developed by LR to highlight Asian ancestry (Fig. S1). Traditional Boolean operators and logical constraints were unreliable or non-functional in both databases; search queries were limited to one or two keywords, and NORD and GARD searches were performed in parallel to identify differences between the two sets of results. All non-duplicate disease reports from NORD and GARD were compiled in Microsoft Excel 2019 version 16.75.2 (23071901) (20) by DI for independent review. Afterward, the remaining rare medical conditions were incorporated into a standardized PubMed search framework to identify scientific literature relevant to the misdiagnosis of each disease (Fig. S2). Authors DI, KA, HS, and LG conducted PubMed searches from October 2021 to January 2023 and DI organized results in Excel for additional independent review (Table S1). To supplement the paucity of epidemiological information about rare diseases, additional references were searched ad-hoc or snowball-sampled from systematically identified PubMed literature. These are noted separately in the results table under the column titled, 'Associated Asian population(s)' (Table 2).

Eligibility criteria and data selection

While reviewing NORD and GARD disease reports, diagnostic disparities were inferred from quantitative and qualitative data of racial and ethnic differences in disease burden, as well as low reported disease prevalence in the U.S. NORD and GARD results were filtered in two rounds of screening: (1) authors KA, DI, and HS removed duplicates and individually marked the remaining results for inclusion or exclusion, and then (2) author LG reviewed all comments to affirm, clarify, or alter prior suggestions about the relevance of each medical condition. This two-step process was repeated while generating and reviewing resultant PubMed studies about the approved diseases.

Eligibility criteria for searches (Table 1) were developed by LG and LR and approved by all authors. During NORD and GARD screening, diseases were accepted for PubMed search queries based on evidence of varied prevalence across racial and ethnic groups, as well as uncommon diagnoses in Asians. For the purposes of this study, a 'rare' disease affects fewer than 200,000 people in the U.S. as per the FDA and Orphan Drug Act (1). Reasons for disease exclusion are shown in Table 1 and Fig. 1. PubMed search results were limited to papers about misdiagnosis in Asian adults published within the last 20 years (i.e. after and including 2003), written or translated into the English language. A maximum of five pieces of eligible literature were recorded per medical condition.

Table 1. Inclusion and exclusion criteria for NORD/GARD disease report screening and PubMed full-text study screening.

Screening round	Inclusion	Exclusion
NORD/GARD screening	The disease is **not commonly** diagnosed in the U.S. because it affects **fewer than** 200,000 people (1).	The disease is **commonly** diagnosed in the U.S. because it affects **more than** 200,000 people.
	The disease demonstrates **varied** prevalence across racial and ethnic groups.	The disease demonstrates **no differences in** prevalence in Asian populations.
		There is **insufficient information** to determine population differences, or there is **no clear relation** to the population(s) of interest.
		The disease **predominantly affects** individuals of **European** descent.
		The disease **predominantly affects children**.
PubMed screening	The paper was published **within the last 20 years.**	The paper was published **more than 20 years ago.**
	The paper primarily discusses (mis)diagnosis in **adult patients.**	The paper primarily discusses diagnosis in **pediatric patients.**
	The paper is **available in English.**	The paper is **not available in English.**

Data extraction and synthesis

Author KA developed a web-parsing code in Python 3.8.11 (21) to conduct iterative searches with LR's search terms. Each search query generated a separate Excel spreadsheet with data columns organized according to the online formatting of NORD and GARD (Table S2). Eligibility decisions were based on information about the racial, ethnic, and geographic distribution of diseases from NORD and GARD; in cases where NORD and GARD provided ambiguous or insufficient descriptions, outside scientific sources were used to facilitate inclusion and exclusion decisions. Eligible PubMed search results were organized in Excel by disease, search date, and article sourcing information.

RESULTS

In our systematic search through the NORD and GARD databases, we found 52 rare diseases that had associations with Asian populations (Fig. 1 and Table 2). This count includes two categories ('Autosomal dominant (spinocerebellar) hereditary ataxia' and 'Gastrointestinal stromal tumors & stomach cancers') representing four distinct NORD and GARD entries ('Autosomal dominant hereditary ataxia', 'Spinocerebellar ataxia 31', 'Gastrointestinal stromal tumors', and 'stomach cancer') that were individually searched in PubMed and then condensed due to etiological and pathological similarity.

The majority (93%) of the initial rare disease search results came from the NORD database, and the remaining from GARD. Broader keywords that encompassed continents ('Asia'), larger regions ('Middle East' and 'Pacific Island'), and race ('Asian') returned relatively more results from NORD and GARD than those describing countries, territories ('Gaza' and 'West Bank'), cities ('Abu Dhabi'), and nationalities/ethnicities ('Chinese', 'Filipino', 'Indian', 'Japanese', and 'Korean'). Some rare conditions were associated with specific Asian countries (e.g. China and Japan) or regions (e.g. Middle East), while others had a general Asian association. Japan, India, and China were the Asian countries with the most associations with the list of rare diseases based on this systematic search. Notably, some Asian populations that were not explicitly searched in NORD/GARD were also implicated in rare disease epidemiology based on descriptions from NORD/GARD and supplemental sources. Examples include blastomycosis in the Vietnamese; leprosy among the Burmese; GNE myopathy in the Persian Jewish; HTLV in Melanesians; sitosterolemia in Micronesians. Out of the 48 total keyword searches that we conducted in the NORD and GARD databases, 12 queries did not generate search results: 'Abu Dhabi', 'Bahrain', 'Bhutan', 'Gaza', 'Indoneasia', 'Jordan', 'Laos', 'Pacific Island', 'Palestine', 'Qatar', 'Timor-leste', and 'West Bank'. However, 'Indonesia' produced 1 NORD entry.

Many of the rare conditions associated with Asian populations are not that rare in certain parts of the world and may even be relatively common in some Asian countries – for example, Behçet's syndrome, cysticercosis, and alpha and beta thalassemias. The rare diseases in our resulting search list include several genetically inherited conditions (e.g. Alport syndrome, Brugada syndrome, CARASIL, and GNE myopathy) and infectious diseases (e.g. blastomycosis, encephalitis, and leishmaniasis). Others are rheumatologic (e.g. Behçet's syndrome), hematologic (e.g. factor XII deficiency), metabolic (e.g. glucose-6-phosphate dehydrogenase deficiency), or oncological (e.g. cholangiocarcinoma).

In the PubMed database, we identified 128 studies that discuss the misdiagnosis and underrecognition of the rare diseases identified in NORD and GARD (Table 2). On average, each rare disease generated 109 PubMed studies within the search framework and two accepted references. There are 11 rare diseases with five accepted references, the maximum number allotted per medical condition in our study. There was a paucity of literature on many of these rare conditions, especially as it related to clinical diagnostics such as misdiagnosis and population demographic considerations for diagnosis. For example, 10 of the 52 conditions yielded zero relevant PubMed search results based on our MeSH search terms alone, either because (1) no results were produced within the search framework ('Autosomal dominant hereditary ataxia', 'Balo disease', 'Banti's syndrome', 'Hemoglobin E disease', and 'Pityriasis rotunda'), or (2) none of the extant research met the PubMed screening criteria ('Encephalitis (West Nile)', 'Factor XII deficiency', 'HTLV Type I and II', 'Sickle cell disease', and 'X-linked dystonia-Parkinsonism/Lubag'). For a given disease, the highest ratio of generated PubMed results to the number of included literature pieces was about 100:1 ('Brucellosis' and 'Filariasis'). Many NORD and GARD entries for the accepted rare diseases also contained minimal to no racial/ethnic distribution data, and additional research was required to further elucidate the epidemiology of affected Asian populations (Table 2).

DISCUSSION

Our systematic analysis and review of the NORD and GARD rare disease databases identified rare diseases that disproportionately impact Asian populations and may be underrecognized and misdiagnosed. These 52 rare diseases span from hereditary conditions such as autosomal dominant hereditary ataxia, to infectious diseases such as melioidosis, to rheumatologic conditions such as Behçet's disease, and to neoplasms such as gastrointestinal stromal tumors and stomach cancers. Although we believe our search efforts were robust, as of the writing of this study, there is a paucity of research and

Table 2. Rare diseases frequently misdiagnosed in Asian patients.

Dx (hyperlinked)	NORD/GARD URL	Dx description	Relevant PubMed literature	Associated Asian* population(s)
Acquired aplastic anemia	https://rarediseases.org/rare-diseases/acquired-aplastic-anemia/	Blood disorder caused by bone marrow failure, often due to autoimmunity.	Iftikhar et al., 2021 (22)	Chinese, Israeli, Malaysian, Thai (23–25)
Alpha thalassemia	https://rarediseases.org/rare-diseases/alpha-thalassemia/	Refers to various genetic blood disorders that cause dysfunctional or absent hemoglobin alpha subunits, which clinically manifest as anemia.	Hamali & Saboor, 2019 (26) Shahid et al., 2017 (27)	Cambodian, Laotian, Malaysian, Middle Eastern (West Asian), Thai
Alport syndrome	https://rarediseases.org/rare-diseases/alport-syndrome/	Genetic disorder associated with chronic kidney disease and ocular-auditory dysfunction. Specific signs and symptoms vary depending on the type of genetic inheritance.	Chen et al., 2021 (28) Wang et al., 2022 (29) Yao et al., 2012 (30) Đurawski et al., 2016 (31)	Middle Eastern (West Asian), Southeast Asian
Autosomal dominant hereditary (spinocerebellar) ataxia	https://rarediseases.org/rare-diseases/autosomal-dominant-hereditary-ataxia/	Genetic neurological disorders characterized by brain/spinal cord degeneration, leading to loss of motor control and speech deficits.	Pedroso et al., 2015 (32)	Chinese, Japanese, Korean, Indian (33–35)
Balo disease (concentric sclerosis)	https://rarediseases.org/rare-diseases/balo-disease/	Rapidly progressive variant of multiple sclerosis caused by demyelination in the brain and spinal cord.		Chinese, Filipino (36)
Banti's syndrome	https://rarediseases.org/rare-diseases/bantis-syndrome/	Premature destruction of red blood cells by the spleen, typically accompanied by splenomegaly and hypertension.		Indian, Japanese (37)
Behçet's syndrome	https://rarediseases.org/rare-diseases/behcets-syndrome/	Idiopathic multi-system inflammatory disorder producing oral and genital ulcers, skin lesions, and ocular abnormalities, among numerous other symptoms.	Sbroglio et al., 2021 (38)	Arabic, Chinese, Iranian, Japanese, Korean, Turkish
Beta thalassemia	https://rarediseases.org/rare-diseases/thalassemia-major/	Genetic blood disorder that presents with low levels of functional hemoglobin and abnormal erythrocytes. The degree of anemia depends on the disease subtype (minor, intermedia, and major).	Islam et al., 2021 (39) Singha et al., 2021 (40) Rizzuto et al., 2021 (41) Rahim et al., 2020 (42)	Cambodian, Chinese, Indian, Middle Eastern (West Asian), South Asian, Southeast Asian (43)
Blastomycosis	https://rarediseases.org/rare-diseases/blastomycosis/	Contagious multisystem fungal infection that can have acute and/or chronic effects. The chronic form disproportionately affects the respiratory and integumentary systems.	Austin et al., 2021 (44) Ballestas et al., 2022 (45) Bethuel et al., 2020 (46) Kumar et al., 2019 (47) Kuzel et al., 2018 (48)	Chinese, Indian, Laotian, Vietnamese (49)
Brucellosis	https://rarediseases.org/rare-diseases/brucellosis/	Bacterial infection that can be transmitted to humans from livestock. The disease can remain localized or evolve with multisystem effects.	Fengzhen et al., 2021 (50) Güven, 2021 (51) Rozis et al., 2021 (52) Yan et al., 2021 (53) Zhai et al., 2021 (54)	East Asian, Middle Eastern (West Asian) (55, 56)
Brugada syndrome	https://rarediseases.org/rare-diseases/brugada-syndrome/	Autosomal dominant genetic disorder that disrupts the cardiac conduction system, producing arrhythmias and sudden death.	Chauveau et al., 2017 (57) Korlipara et al., 2021 (58) Smith et al., 2021 (59)	Japanese, Middle Eastern (West Asian), Thai (60)
Buerger's disease (thromboangiitis obliterans)	https://rarediseases.org/rare-diseases/buergers-disease/	The occlusion of blood vessels in the extremities, most often in tobacco users. In its most severe form, it can lead to gangrene.	Baran et al., 2017 (61)	Indian, Israeli, Japanese, Korean, Middle East (West Asian) (62)
CARASIL	https://rarediseases.org/rare-diseases/carasil/	A genetically inherited form of cerebral subcortical arteriopathy that destroys myelin in the brain and spinal cord. Patients frequently experience stroke-like episodes with speech and motor deficits.	Bersano et al., 2021 (63) Kitahara et al., 2022 (64)	Chinese, Japanese (65, 66)
Cerebrotendinous xanthomatosis	https://rarediseases.org/rare-diseases/cerebrotendinous-xanthomatosis/	Autosomal recessive disorder causing cholesterol deposits in the nervous system, eyes, arteries, and tendons. Often associated with CHD, seizures, cognitive impairment, and ataxia.	Bajaj et al., 2013 (67) Cao et al., 2020 (68) Koyama et al., 2021 (69) Lee et al., 2019 (70) Sekijima et al., 2018 (71)	Chinese, Japanese, Iranian, Israeli (72, 73)
Chikungunya	https://rarediseases.org/rare-diseases/chikungunya/	Viral infection from mosquito bites that causes rash, fever, and severe joint pain (arthritic virus).	Cotella et al., 2021 (74) Ferede et al., 2021 (75) Hakami et al., 2021 (76) Mala et al., 2021 (77) Stubbs et al., 2020 (78)	Indian, Oceanian, Southeast Asian (79, 80)

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Table 2. (Continued)

Dx (hyperlinked)	NORD/GARD URL	Dx description	Relevant PubMed literature	Associated Asian* population(s)
Cholangiocarcinoma	https://rarediseases.org/rare-diseases/cholangiocarcinoma/	Bile duct cancers that can originate at various points inside or near the liver and its openings.	Liu et al., 2021 (81)	Indian
Cronkhite-Canada syndrome	https://rarediseases.org/rare-diseases/cronkhite-canada-syndrome/	Acquired intestinal disease characterized by polyps, ageusia, and abnormal nail growth.	Fan et al., 2016 (82) Slavik & Montgomery, 2014 (83)	Chinese, Japanese (84)
Cysticercosis	https://rarediseases.org/rare-diseases/cysticercosis/	Parasitic infection that can cause cysts in various parts of the body, often the nervous system and eyes.	Cantey et al., 2021 (85) Gupta et al., 2021 (86) Liu et al., 2021 (81) Lobo et al., 2021 (87) Wankhede et al., 2021 (88)	South Asian, Southeast Asian (89)
Dengue fever	https://rarediseases.org/rare-diseases/dengue-fever/	Acute viral infection transmitted by mosquito bites, with symptoms ranging from mild fever to fatal hemorrhage.	Jisamerin et al., 2021 (90) Umakanth & Suganthan, 2020 (91) Puthalath et al., 2021 (92)	Oceanian, South Asian, Southeast Asian (93–95)
Diffuse panbronchiolitis	https://rarediseases.org/gard-rare-disease/diffuse-panbronchiolitis/	Bronchiolitis coupled with chronic sinusitis that commonly presents with productive cough, shortness of breath, and drastic weight loss. Can quickly evolve to bronchiectasis, respiratory failure, and death.	Atici et al., 2005 (96) Chen et al., 2005 (97)	Japanese
Elephantiasis	https://rarediseases.org/rare-diseases/elephantiasis/	Gross enlargement of body parts due to excess lymph (severe lymphedema). Possible causes include lymphatic filariasis, STDs, tuberculosis, leishmaniasis, leprosy, and other bacterial infections.	Murthy, 2019 (98)	Indian
Encephalitis (Japanese)	https://rarediseases.org/rare-diseases/encephalitis-japanese/	Severe brain inflammation/damage caused by infection of Japanese B encephalitis virus, transmitted by mosquitoes.	Bharucha et al., 2018 (99) Hotez et al., 2015 (100) Tarantola et al., 2014 (101)	Japanese
Encephalitis (West Nile)	https://rarediseases.org/rare-diseases/west-nile-encephalitis/	(A)symptomatic illness caused by West Nile encephalitis virus, transmitted by mosquitoes.		East Asian, Middle Eastern (West Asian), South Asian (102)
Esophageal cancer	https://rarediseases.org/rare-diseases/esophageal-cancer/	Malignant cancers of the esophagus, typically originating in mucosal cells. The main types include squamous cell carcinoma and adenocarcinoma.	Ahmadi et al., 2020 (103) Kimura et al., 2020 (104) Nobel et al., 2020 (105) Visaggi et al., 2021 (106)	East Asian, Middle Eastern (West Asian) (107)
Factor XII deficiency	https://rarediseases.org/rare-diseases/factor-xii-deficiency/	Asymptomatic genetic blood disorder causing prolonged blood clotting due to deficiency of Hageman factor (XII). Typically does not cause a predisposition to prolonged bleeding, making it overall benign.		East Asian (108, 109)
Fascioliasis	https://rarediseases.org/rare-diseases/fascioliasis/	Parasitic liver infection from water-growing plants. Includes Halzoun Syndrome, a variant of fascioliasis that affects the pharynx.	Ahmad et al., 2021 (110) Caravedo & Cabada, 2020 (111) Dermauw et al., 2021 (112) Lee et al., 2021 (113) Siles-Lucas et al., 2021 (114)	Chinese, Egyptian, Japanese, Iranian, Oceanian, Vietnamese, Turkish (115, 116)
Filariasis	https://rarediseases.org/rare-diseases/filariasis/	Parasitic infection transmitted by mosquitoes that causes fever, lesions, and chronic lymphatic obstruction.	Mitchell et al., 2022 (117) Ngatse et al., 2022 (118)	Chinese, Indian, Southeast Asian (119, 120)
Gastrointestinal stromal tumors (GISTs) and stomach cancers	https://rarediseases.org/rare-diseases/gastrointestinal-stromal-tumors/	Malignant sarcomas that can originate in nerve cells anywhere along the gastrointestinal tract, with most occurring in the stomach or small intestine. Pathologically distinct from mycobacterial infections that cause tuberculosis and leprosy.	Chadwick et al., 2015 (121) Quinones et al., 2023 (122) Sung et al., 2011 (123) Tata et al., 2013 (124) Zheng et al., 2022 (125)	Chinese, Korean (126)
Glucose-6-phosphate dehydrogenase deficiency	https://rarediseases.org/rare-diseases/glucose-6-phosphate-dehydrogenase-deficiency/	Genetic metabolic disease that can be conditionally triggered to cause premature hemolysis, which clinically manifests as anemia.	C. Albayrak & D. Albayrak, 2015 (127) Palmer et al., 2020 (128) Tang et al., 2017 (129) Phillpotts et al., 2014 (130)	Chinese, Indian, Southeast Asian (131)

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Table 2. (Continued)

Dx (hyperlinked)	NORD/GARD URL	Dx description	Relevant PubMed literature	Associated Asian* population(s)
GNE Myopathy (HIBM, Nonaka myopathy, IBM2, etc.)	https://rarediseases.org/rare-diseases/gne-myopathy/	Autosomal recessive musculoskeletal disease causing progressive muscle weakness.	Awasthi et al., 2019 (132) Soule et al., 2018 (133)	Japanese, Persian Jewish
Hemoglobin E disease	https://rarediseases.org/gard-rare-disease/hemoglobin-e-disease/	Autosomal recessive blood disorder characterized by misshapen hemoglobin E and mild anemia.		Chinese, Sri Lankan, Thai (134, 135)
HTLV (Type I, II)	https://rarediseases.org/rare-diseases/htlv-type-i-and-type-ii/	Infection with HTLV-1 virus, which can evolve into a chronic neurologic disease that can take the form of acute T-cell leukemia/lymphoma (ATL) or HTLV-I associated myelopathy/tropical spastic paraparesis (HAM/TSP).		Japanese, Melanesian, Middle Eastern (West Asian)
Kennedy disease	https://rarediseases.org/rare-diseases/kennedy-disease/	X-linked neuromuscular disorder that primarily causes progressive muscle weakness and dysphagia.	Harutunian et al., 2013 (136) Paparounas et al., 2003 (137) Skoretz et al., 2012 (138) Yuan et al., 2016 (139)	Japanese
Keratomalacia	https://rarediseases.org/rare-diseases/keratomalacia/	Ocular disease caused by severe vitamin A deficiency, frequently compromising vision corneal texture (keratomalacia) bilaterally. Can evolve with gray deposits on the whites of the eyes (Bitot spots), ocular infection, rupture, and blindness.	Lai et al., 2014 (140)	South Asian, Southeast Asian (141, 142)
Kikuchi's disease (histiocytic necrotizing lymphadenopathy)	https://rarediseases.org/rare-diseases/kikuchis-disease/	Benign lymph cancer causing enlarged, inflamed lymph nodes.	Cunha et al., 2009 (143) Mirgh et al., 2016 (144) Murali et al., 2014 (145) Rakesh et al., 2014 (146) Schofer et al., 2005 (147)	Japanese, Southeast Asian (148)
Kimura disease	https://rarediseases.org/gard-rare-disease/kimura-disease/	Idiopathic, noncancerous subcutaneous masses that appear in head and neck tissue. Also referred to as eosinophilic (lympho) granuloma and eosinophilic lymphofolliculosis, among many other names.	Gong et al., 2015 (149) Ye et al., 2015 (150) Zhang & Jiao, 2019 (151)	Chinese (152)
Leishmaniasis	https://rarediseases.org/rare-diseases/leishmaniasis/	Contagious parasitic infection transmitted by sand flies, which can manifest as cutaneous, mucosal, or visceral. The disease may present asymptomatically or with moderate to life-threatening complications.	Andrade et al., 2022 (153) Behniafar et al., 2021 (154) Ikedionwu et al., 2021 (155) Volpedo et al., 2021 (156)	Bangladeshi, Indian, Middle Eastern (West Asian), Nepalese
Leprosy	https://rarediseases.org/rare-diseases/leprosy/	Infectious human disease of the skin, mucosal membranes, and peripheral nerves that can lead to muscle paralysis, disfigurement, and blindness. Presents as tuberculoid (mild, localized), borderline (intermediate), or lepromatous (severe, diffuse).	Bernardes-Filho et al., 2021 (157) Sinha et al., 2021 (158) Swain et al., 2022 (159)	Bangladeshi, Burmese, Indian, Indonesian
Melioidosis	https://rarediseases.org/gard-rare-disease/melioidosis/	Bacterial infection obtained from contaminated soil and water, with a similar clinical presentation to tuberculosis and pneumonia.	Chatterjee et al., 2021 (160) Goarant et al., 2021 (161) Gopalakrishnan, 2021 (162) Veluthat et al., 2021 (163) Virk et al., 2020 (164)	South Asian, Southeast Asian (165)
Metachromatic leukodystrophy	https://rarediseases.org/rare-diseases/metachromatic-leukodystrophy/	Genetic neurodegenerative disease causing demyelination throughout the nervous system. All subtypes (late-infantile, juvenile, adult) cause intellectual, cognitive, and motor dysfunction.	Pekgül et al., 2020 (166) Politi et al., 2018 (167)	Middle Eastern (West Asian)
Miller Fisher syndrome	https://rarediseases.org/rare-diseases/miller-fisher-syndrome/	Acquired, acute autoimmune nerve disease that manifests after bacterial/viral infection, similar to Guillain-Barré syndrome. Presents with temporary ocular and motor dysfunction.	Martins et al., 2020 (168) Zeylikman et al., 2015 (169)	Japanese (170)
Moyamoya disease	https://rarediseases.org/rare-diseases/moyamoya-disease/	Progressive cerebrovascular disease causing simultaneous carotid artery occlusion and small 'moyamoya' vessel vasodilation. Presents like TIA or CVA (stroke). Disease can be acquired genetically, secondary to other concurrent disease or idiopathically.	Koul et al., 2021 (171) Ma et al., 2019 (172) Mohammadi et al., 2019 (173) Musa et al., 2021 (174) Sapra et al., 2020 (175)	Japanese

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Table 2. (Continued)

Dx (hyperlinked)	NORD/GARD URL	Dx description	Relevant PubMed literature	Associated Asian* population(s)
Nontuberculous mycobacterial lung disease	https://rarediseases.org/rare-diseases/nontuberculous-mycobacterial-lung-disease/	Refers to acute, acquired mycobacterial infections that do not cause tuberculosis or leprosy. Symptoms vary greatly across patients and organ systems, but the disease can become chronic.	Doyle et al., 2020 (176) Liu et al., 2022 (177)	Chinese, Filipino, Japanese, Korean, Vietnamese (178, 179)
Palmoplantar pustulosis	https://rarediseases.org/rare-diseases/palmoplantar-pustulosis/	Chronic, idiopathic inflammatory disorder presenting with painful, yellow blisters on hands and feet.	Huang & Tsai, 2020 (180) Su et al., 2011 (181)	Japanese
Pityriasis rotunda	https://rarediseases.org/gard-rare-disease/pityriasis-rotunda/	Skin disease causing round, scaly, pigmented patches on the trunk and limbs. Type 1 is malignant; type 2 is benign.		Japanese (182, 183)
Sickle cell disease	https://rarediseases.org/rare-diseases/sickle-cell-disease/	Autosomal recessive blood disorder causing crescent-shaped, stiff red blood cells that inhibit circulation (anemia). Left untreated, it can evolve to multisystem organ damage and failure.		Indian, Middle Eastern (West Asian)
Sitosterolemia	https://rarediseases.org/rare-diseases/sitosterolemia/	Autosomal recessive disease that prevents excretion of plant sterols.	Bastida et al., 2019 (184) Bazerbachi et al., 2017 (185) Okafor et al., 2022 (186)	Chinese, Japanese, Indian, Micronesian
Tropical sprue	https://rarediseases.org/rare-diseases/tropical-sprue/	Idiopathic digestive disease causing malabsorption in the small intestine and nutritional deficiencies.	Brown et al., 2014 (187)	Indian, Southeast Asian
Tularemia	https://rarediseases.org/rare-diseases/tularemia/	Bacterial infection transmitted by small mammals and arthropods. Most cases begin with rapid onset of flu-like symptoms, but subsequent symptoms vary across patients and affected organ systems.	Atmaca et al., 2009 (188) Calanan et al., 2010 (189) Donate-Pérez-Molino et al., 2019 (190) Maurin, 2020 (191) Sateia et al., 2017 (192)	Japanese, Turkish (193)
Typhoid	https://rarediseases.org/rare-diseases/typhoid/	Bacterial infection causing fever, weight loss, abnormal bowel movements, and internal bleeding, among other symptoms. Patients can be carriers long after recovery.	Esa et al., 2021 (194)	Chinese, Indian, Indonesian, Malaysian, Nepalese, Pakistani, Vietnamese (195–197)
Vogt-Koyanagi-Harada disease	https://rarediseases.org/gard-rare-disease/vogt-koyanagi-harada-disease/	Idiopathic ocular, auditory, nerve, and musculoskeletal disease that presents with vitiligo, alopecia, cataracts, and glaucoma.	Shoughy & Tabbara, 2019 (198) Yang et al., 2018 (199)	Chinese, Indian, Japanese, Thai (200, 201)
X-linked dystonia-parkinsonism (Lubag)	https://rarediseases.org/gard-rare-disease/x-linked-dystonia-parkinsonism-lubag/	Neurodegenerative disorder causing unintended motor movements as a result of adult-onset Parkinsonism and/or focal dystonia.		Filipino (202)

*** 'Asian'** is defined as people with ancestries from *East Asia* (China, Japan, Korea, Mongolia); *South Asia* (Bangladesh, Bhutan, India, Nepal, Sri Lanka); *Southeast Asia* (Cambodia, Indonesia, Laos, Malaysia, Philippines, Singapore, Thailand, Timor-leste, Vietnam); *West Asia and the Middle East* (Afghanistan, Bahrain, Egypt, Iraq, Israel, Jordan, Kuwait, Lebanon, Oman, Pakistan, Palestine, Qatar, Saudi Arabia, Syria, Turkey, United Arab Emirates, Yemen); *Oceania and the greater Pacific Islands* (Melanesia, Micronesia, Polynesia). *Melanesia* includes Fiji, New Caledonia, Papua New Guinea, Solomon Islands, and Vanuatu. *Micronesia* includes the Federated States of Micronesia, Guam, Kiribati, Marshall Islands, Nauru, Northern Mariana Islands, and Palau.

medical education regarding rare diseases, particularly for these conditions that disproportionately affect individuals of Asian descent.

While the exact reasons for the differences in the prevalence of rare diseases among Asian populations are not fully understood (nor are they within the scope of this review), it is likely that genetic, environmental, demographic, geographic, and sociocultural factors exert varying and manifold influence in some or many of the conditions. Many rare diseases are caused by genetic mutations. As another example, certain genetic mutations might be more common in specific populations due to ancestral heritage and geographic distributions of these populations (203–206). Furthermore, cultural practices such as consanguineous marriage are associated with increased rates of certain inherited genetic conditions, particularly autosomal recessive conditions (207). For instance, diseases such as type 2 diabetes mellitus; breast, colorectal, prostate, and lung cancers; anemia, thalassemia, and cardiovascular diseases; as well as

hearing deficits are known to be associated with consanguinity in Asian countries (208, 209). Additionally, environmental factors can influence the development of rare diseases (210, 211). Exposure to various toxins, chemicals, infections, or dietary elements may heighten the risk of specific rare diseases, creating differences in their prevalence across populations (212, 213). For instance, the consumption of pickled vegetables and salted fish has been shown to be associated with onset of gastric cancer in Asian populations (214, 215).

A major challenge we found in conducting this study is the paucity of data and literature in this area, which may be due to several reasons. One primary reason is the relatively lower prevalence of these diseases in Western populations, resulting in limited awareness, focus, and funding for such research. Unfortunately, some of these conditions are recognized as blind spots and labeled as 'neglected' conditions (216). There is also relatively lower funding and resources for rare diseases in general, which is a challenge to advance the collective knowledge and care for patients suffering from these conditions (217). Additionally, cultural and language barriers pose significant obstacles, hindering the dissemination of information and collaboration between researchers from different regions (218). Moreover, some Asian cultures associate a stigma with certain health conditions, leading patients to be hesitant in disclosing their symptoms or seeking medical help due to fear of judgment or discrimination (219).

The PubMed search results demonstrate that many of these conditions are susceptible to misdiagnosis (Table 2). Misdiagnoses are a critical concern as they can lead to delayed or ineffective treatments, exacerbating the suffering of patients and hindering efforts to maintain functionality, obtain a cure, recover, or otherwise achieve effective disease management. It is imperative for healthcare institutions, funding agencies, and researchers worldwide to prioritize strategies and studies to address misdiagnosis and health disparities among diverse populations. Collaborative efforts between medical centers in Western countries and those in Asian regions could facilitate information exchange, enhance research capacities, and promote more inclusive clinical trials and studies. Moreover, raising awareness among the general public and healthcare providers about the prevalence and specific manifestations of these rare diseases in Asian populations is paramount. This can lead to earlier diagnosis, appropriate treatment, and better patient outcomes.

Our study had several limitations, including database limits, record keeping practices, and varying country-by-country case definitions for rare diseases. NORD and GARD provide information on many rare diseases but do not index all rare diseases. While there are estimated to be more than 10,000 rare diseases globally, NORD and GARD only index 1,200 and 7,000, respectively (2, 19, 220). The results are limited by the search terms used in the study, and we may have missed certain relevant rare diseases in the databases. For example, infections such as tuberculosis (221) and strongyloidiasis (222) and rheumatologic conditions such as IgG4-related disease (223), Familial Mediterranean Fever (224, 225), and Takayasu arteritis (226) may disproportionately impact Asian populations depending on exposures and sociodemographic risk factors. Other limitations include the variable standards and reliability of record keeping of rare diseases within other countries (227). The recognition, diagnosis, and recording practices across different countries and health systems can also impact the reported prevalence of rare diseases. The majority of the data in this study came from China, Japan, and India, potentially limiting the study's breadth. In regions where medical professionals are more familiar with certain rare conditions, they may diagnose them more frequently, leading to apparent higher prevalence in those areas. A condition may be more prevalent in reality than it appears in the literature but may not be well-captured by diagnosis or recording mechanisms. Additionally, our search strategy was conducted in English, and, therefore, publications in other languages may not have been captured well. Moreover, limited access to healthcare facilities and medical expertise in certain regions can affect the diagnosis and reporting of rare diseases, potentially underestimating their true prevalence (228). Studying the factors contributing to these differences can lead to improved diagnosis, treatment, and prevention strategies for affected individuals worldwide. This study was also limited by the exclusion of the pediatric conditions that constitute a significant population of rare diseases, especially genetically inherited and congenital conditions.

This study is the first to systematically analyze rare disease databases to identify rare conditions in the U.S., which disproportionately affect Asian populations and review their potential for misdiagnosis. We also underscore and review the disparities and misdiagnoses that have been reported to occur among Asian patients. This report adds to the paucity of literature in the subject of rare diseases, especially for diverse populations, and can inform medical curricula development and health professional training to improve Asian health outcomes.

The findings of this study underscore the pressing need for enhanced awareness, research, and medical education concerning rare diseases in Asian communities. It is vital for healthcare professionals, particularly those practicing in regions with significant Asian populations, to be equipped with the knowledge and resources essential for accurate recognition and diagnosis of these conditions. Furthermore, our work will serve as a crucial source of information to inform precision medicine techniques for diagnosing rare diseases in Asian populations. By providing nuanced

insights into clinical diagnoses, this research lays the foundation for improving education and ultimately enhancing health outcomes for Asian individuals, and by extrapolation of methods, the health status of all peoples. In recognizing that Asian patients come from diverse backgrounds and possess specific needs, precision medicine offers a powerful means of personalizing healthcare. By tailoring treatments to the individual genetic, cultural, geographic, and environmental exposures and profiles of Asian patients, we can avoid the pitfalls of one-size-fits-all approaches and better ensure the delivery of the most suitable and effective evidence-based care.

Future directions and next steps of this work hold immense promise in advancing our understanding and support for individuals with rare disorders. Drawing on the valuable insights from this scoping review, researchers should direct their attention to the diagnostic barriers of rare diseases in Asian populations, with a specific focus on the influential factors. These factors encompass the impact of culture, language, medical provider and patient education, economic and financial considerations, as well as the role of social determinants of health. By thoroughly examining how these elements interplay, researchers can better understand and address the challenges that influence health outcomes in Asian populations affected by rare diseases. Additionally, while the current work focused on the recognition of rare diseases in adult Asian populations toward improved diagnosis, future research should expand these investigations to include pediatric and other racial and ethnic populations.

ARTICLE INFORMATION

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Data availability statement

The source data were derived from the following database resources available in the public domain: National Organization for Rare Disorders (NORD) at https://rarediseases.org/ and Genetic and Rare Diseases Information Center (GARD) at https://rarediseases.info.nih.gov/. The web-parsing search algorithm developed in Python is available on GitHub at https://github.com/ihdaph/CARE21RareDx.

Conflict of interest and funding

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