DRAFT COMPETENCY STANDARDS AND KNOWLEDGE GUIDANCE

to support health workers' diagnosis of rare diseases

CONTENTS

INTRODUCTION

This document outlines the draft competency standards for health workers seeking to make a diagnosis when a person presents with evolving, vague or unexplained symptoms. The challenge for the clinician in these settings is to distinguish common conditions from rare diseases. Most presentations with vague or unexplained symptoms are not rare diseases; but a proportion of those that have repeated presentations for these symptoms will have a rare disease, and they may experience significant delays to diagnosis.

The standards focus on the period before diagnosis, and highlight the responsibility of clinicians to listen, reflect, seek advice and collaborate with the patient. Some rare diseases – for example, severe combined immunodeficiency – are marked by a first presentation in crisis, and the diagnosis is often made through pattern recognition and tests performed for a severely ill person. Many rare diseases present with polymorphous symptoms such as fatigue, weakness, or respiratory symptoms, and these may take considerable endeavour to diagnose. Some symptom clusters may be classified as medically undiagnosed; the diagnostic journey for these patients requires as much patience and respectful collaboration as the journey to the diagnosis of a rare disease.

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The competencies are organised under the six domains used in the World Health Organization's Global Competency Framework for Universal Health Coverage[1]:

- Domain I   People-centredness
- Domain II   Decision-making
- Domain III   Communication
- Domain IV   Collaboration
- Domain V   Evidence-informed practice
- Domain VI   Personal conduct

For each competency standard a set of behaviours are listed which demonstrate how the competency would be delivered by health workers. The knowledge, skills and attitudes constitute the knowledge guide for designers of education programs.

These competency standards are not focused around the acquisition of knowledge about rare diseases. We assume that most first-contact clinicians should not aim for an encyclopaedic knowledge of rare diseases, but rather be prepared to countenance the possibility that the client may have one, if their symptoms are long standing or relapsing and do not fit the working

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[1] Global competency framework for universal health coverage. Geneva: World Health Organization; 2022 (https://apps.who.int/iris/handle/10665/352710).

diagnosis. Patients frequently have researched the possible diagnosis of a rare disease, and may be more knowledgeable than their clinicians.

We encourage doctors to share the diagnostic journey with their patients. Not all diagnostic journeys end in the diagnosis of a rare disease, but all are enriched by the respectful collaboration of patient and doctor.

DOMAIN 1: People-centredness

Competency standard 1:

Provides people-centred care that is sensitive, respectful and adapted to individual needs

Behaviours	1.1 Provides care that recognises the physical, social and emotional impacts on symptoms of illness	**Knowledge** • Outlines the physical, social and emotional impacts of undiagnosed rare diseases and/or medically unexplained symptoms[2]
		Skills • Demonstrates integration of psychosocial and emotional impacts into care for individuals with evolving and/or medically unexplained symptoms
		Attitudes • Seeks to understand the holistic impacts of undiagnosed rare diseases and/or medically unexplained symptoms
	1.2 Adopts an approach to practice that is non-blaming, non-discriminatory, non-judgemental and non-stigmatizing	**Knowledge** • Articulates the harms that may result from failure to provide respectful care to individuals in the case of evolving and/or medically unexplained symptoms
		Skills • Identifies and seeks to mitigate stigmatisation and discrimination that may be faced by individuals with evolving and/or medically unexplained symptoms
		Attitudes • Supports non-judgemental access to care for people with undiagnosed rare diseases and/or medically unexplained symptoms
	1.3 Demonstrates sensitivity to cultural, social and gender impacts and expressions of illness	**Knowledge** • Outlines gendered presentations of selected rare diseases • Outlines social and cultural impacts of selected rare diseases

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[2] Husain M, Chalder T. Medically unexplained symptoms: assessment and management. *Clin Med (Lond)*. 2021;21(1):13-18.

		Skills • Applies a gender, social or cultural lens to analyse evolving and medically unexplained symptoms
		Attitudes • Engages with interest in the social and cultural context of the individual

Competency standard 2:

Promotes the agency of the individual in health care settings

Behaviours	2.1 Promotes confidence in individuals to seek care for their symptoms.	Knowledge • Outlines importance of individual agency in health outcomes
		Skills • Demonstrates ability to support confidence and agency in individuals • Solicits and responds to individual's observations and interpretations of their own symptoms
		Attitudes • Prioritises the contributions and self-assessments of individuals with evolving or medically unexplained symptoms
	2.2 Promotes health literacy	Knowledge • Outlines components of health literacy,[3] including digital health literacy[4]
		Skills • Demonstrates strategies to support an individual's health literacy in relation to rare diseases, evolving and/or undiagnosed medical symptoms
		Attitudes • Demonstrates ability to promote health literacy • Values learning from patients

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[3] Australian Commission on Safety and Quality in Health Care. Health literacy: Taking action to improve safety and quality. Sydney: ACSQHC, 2014.
[4] Four components of digital health literacy: Paige SR, Stellefson M, Krieger JL, et al. Proposing a transactional model of eHealth literacy: concept analysis. *J Med Internet Res*. 2018;20(10):e10175.

DOMAIN II: Decision-making

> **Competency standard 3:**
>
> Takes a collaborative and adaptive approach to decision-making in evolving situations

Behaviours	3.1 Promotes adaptive and collaborative decision-making in conditions of uncertainty	Knowledge • Outlines elements of shared decision-making
		Skills • Works collaboratively with individuals to set and review goals and actions in conditions of uncertainty
		Attitudes • Values the contribution of the individual to decision-making
	3.2 Reassesses working diagnoses as symptoms of illness change.	Knowledge • Identifies "red flags" for rare diseases[5]
		Skills • Seeks out "red flags" for rare diseases that would suggest reassessment of working diagnoses of evolving and/or medically unexplained symptoms.
		Attitudes • Maintains openness to new or different diagnoses

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[5] The Family GENES mnemonic sums up the Red Flags: Family history; groups of anomalies; Early or Extreme presentations of common diseases, Neurodevelopmental or Neurodegenerative conditions; Exceptional or unusual pathology; and Surprising laboratory values. Whelan AJ, Ball S, Best L, et al. Genetic red flags: clues to thinking genetically in primary care practice. *Prim Care* 2004;31:497-508.

The National Recommendations for Rare Disease Health Care (2024) adds to this: Multiple visits to different specialists without a diagnosis.

DOMAIN III: Communication

> **Competency standard 4:**
>
> Communicates with clarity and sensitivity in accordance with the needs and purpose of the interaction

Behaviours	4.1 Adapts communication to changing context, illness settings and events.	**Knowledge** • Outlines situations where interpreters should be engaged
		Skills • Adapts communication style to need and setting • When needed, demonstrates ability to communicate respectfully with family members • Uses clear, non-technical language • Works effectively with interpreters
		Attitudes • Responds receptively to changing circumstances and illness concerns • Respects the confidentiality of the individual
	4.2 Acknowledges limits of knowledge.	**Knowledge** • Identifies limitations in knowledge about diagnoses for rare diseases
		Skills • Communicates effectively about grey areas of medical knowledge • Communicates effectively about the limitations of one's own knowledge and experience
		Attitudes • Maintains a non-judgemental approach to individuals seeking a diagnosis • Recognises the limits of one's own knowledge
	4.3 Exchanges information to support shared goal-setting.	**Knowledge** • Outlines communication approaches to be used to drive shared decision-making [6]
		Skills • Undertakes shared decision-making in relation to evolving and/or medically unexplained symptoms
		Attitudes • Respects the knowledge of the individual • Supports self-efficacy

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[6] The SHARE Approach: a model for shared decision-making. Agency for Healthcare Research and Quality, Rockville MD. September 2020 https://www.ahq.gov/health-literacy/professional-training/shared-decision/tools;factsheet.html; Légaré F, Witteman HO. Shared decision making: examining key elements and barriers to adoption into routine clinical practice. Health Aff (Millwood). 2013 Feb;32(2):276-84. doi: 10.1377/hlthaff.2012.1078. PMID: 23381520.

DOMAIN IV: Collaboration

Competency standard 5:

Collaborates purposefully with health workers for individuals with evolving or unexplained illnesses

Behaviours	5.1 Engages in collaborative practice with other health workers in evolving clinical conditions.	Knowledge • Clarifies benefits of working collaboratively with other health workers • Identifies health workers with relevant experience for the evolving condition, or suspected rare disease
		Skills • Works collaboratively with other health workers in the care of people with evolving and/or medically unexplained symptoms.
		Attitudes • Appreciates the diversity of skills of other health workers • Values second opinions

DOMAIN V: Evidence-informed practice

> **Competency standard 6:**
>
> Uses evidence to inform the care of individuals with evolving or unexplained illnesses.

Behaviours	6.1: Applies principles of evidence-informed practice in investigation of symptoms.	**Knowledge** • Identifies a range of evidence-informed tools to inform investigation
		Skills • Applies evidence-informed guidelines to the challenges of diagnosis, including over-investigation[7]
		Attitudes • Values evidence-informed practice
	6.2: Assesses data and information from a range of sources.	**Knowledge** • Describes evidence-informed sources of information about rare diseases and medically unexplained illnesses
		Skills • Critically analyses and applies data from different sources to the task of diagnosis • Refers people to reliable sources of information
		Attitudes • Values and supports reliable information and data
	6.3: Identifies and discusses misinformation about symptoms of medically unexplained and/or diagnoses of rare diseases.	**Knowledge** • Defines misinformation[8]
		Skills • Identifies examples of misinformation in relation to evolving and/or medically unexplained symptoms • Counters and respectfully challenges misinformation [9]
		Attitudes • Advocates against the use of misinformation

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[7] Hoffmann TC, Del Mar C. Patients' expectations of the benefits and harms of treatments, screening, and tests: a systematic review. JAMA Intern Med. 2015 Feb;175(2):274-86.

[8] Office of the Surgeon General (OSG). Confronting Health Misinformation: The U.S. Surgeon General's Advisory on Building a Healthy Information Environment. Washington (DC): US Department of Health and Human Services; 2021.

[9] Shajahan A, Pasquetto IV. Countering Medical Misinformation Online and in the Clinic. Am Fam Physician. 2022 Aug;106(2):124-125.

DOMAIN VI: Personal conduct

Competency standard 7:

Works within the limits of competence and scope of practice

Behaviours	7.1: Provides care and advice in accordance with the scope of practice of the discipline.	Knowledge • Outlines the scope of practice of a discipline in relation to care of unexplained symptoms and diagnosis of evolving illnesses. • Outlines the determinants of competence in caring for unexplained symptoms or evolving illnesses.
		Skills • Responds to unexplained symptoms and evolving illnesses within scope of discipline. • Demonstrates knowledge of one's own competence in relation to evolving and/or medically unexplained symptoms.
		Attitudes • Demonstrates respect for health care disciplines other than one's own • Recognises the limits of one's own knowledge and skills

Competency standard 8:

Engages in ongoing learning and reflective practice

Behaviours	8.1: Engages in ongoing learning of relevance to unexplained or evolving illnesses.	Knowledge • Advocates for continuing education in relation to rare diseases, and care for medically unexplained symptoms
		Skills • Assesses ongoing learning needs in relation to rare diseases and management of medically unexplained symptoms • Undertakes continuing education about rare diseases and their presentations
		Attitudes • Values ongoing education about rare diseases and challenging diagnoses

	8.2 Reflects upon and addresses one's own attitudes about medically unexplained symptoms	Knowledge • Explains the concept of reflexivity[10]
		Skills • Applies reflexivity to one's own clinical practice
		Attitudes • Recognises one's own cultural, gender or social biases

[10] Olmos-Vega FM, Stalmeijer RE, Varpio L, Kahlke R (2023) A practical guide to reflexivity in qualitative research: AMEE Guide No. 149, Medical Teacher, 45:3, 241-251, DOI: 10.1080/0142159X.2022.2057287