Rare Diseases of Public Health Significance

Most nationally notifiable communicable conditions are specific infections such as salmonellosis or pertussis. However, Washington also has a category of reportable conditions that are rarely seen in the state or are less well understood compared to more common conditions. The category of Rare Disease of Public Health Significance enables reporting of new or unusual conditions that could impact the state’s residents.

Notifiable Conditions Reporting

Public health agencies establish reporting of diseases in order to take action. There may be a risk of transmission from a case patient, the need to identify a source of exposure, or an opportunity to identify an unusual condition that does not typically occur in Washington. While known conditions can be specified for reporting and the associated public health actions defined, new or emerging conditions may also be of public health importance and of national concern.

Under Washington Administrative Code (WAC) 246-101, there are specific reporting requirements for healthcare providers and healthcare facilities to report notifiable diseases. Providers and facilities are also asked to report cases or clusters of rare diseases of public health significance, defined in the WAC as:

“… a disease or condition, of general or international public health concern, which is occasionally or not ordinarily seen in the state of Washington including, but not limited to, spotted fever rickettsiosis, babesiosis, tick paralysis, anaplasmosis, and other tick borne diseases. This also includes public health events of international concern and communicable diseases that would be of general public concern if detected in Washington.”
This definition specifies conditions that were of interest at the time the notifiable conditions WAC was last revised in 2011 as well as outlining a broader category of unusual diseases or events with potential public health implications. There are several purposes for asking that providers report these rare diseases.

Reported cases help outline the epidemiology of emerging and uncommon diseases in Washington State residents and may be a public health early warning system. Documenting the presence of an infectious agent in the state can increase provider awareness of the agent and therefore the likelihood of identifying additional cases. For example, coccidioidomycosis (Valley fever) was not known to be acquired in the state until 2010-2011 when several cases were investigated and local exposures identified in central Washington. There has been outreach to providers to consider the diagnosis of coccidioidomycosis even without travel to the more prominent endemic areas of southwest Washington or Central America.

For some rare diseases, commercial testing may be unavailable or not readily available. Public health agencies may be able to arrange testing at a state or national reference laboratory such as at the Washington State Department of Health or the Centers for Disease Control and Prevention. Case treatment and management recommendations may also be available from these sources. A disease such as human rabies is extremely rare. If a provider suspects a case, prompt diagnostic support can be offered at the public health reference laboratory and resources for treatment protocols provided.

Rare diseases that are communicable may result in variable public health intervention. It may be appropriate to identify potentially exposed persons who shared an exposure, or who were exposed to the patient or specimens; included may be household contacts, laboratory workers handling a culture, or even healthcare providers. For certain agents, public health actions may be initiated even before an agent is confirmed. A list of contacts might be developed for a suspected case of a newly emerging severe coronavirus infection like MERS, with the potential for transmission in healthcare settings.
The investigation of any notifiable condition usually involves attempting to identify sources of transmission and as appropriate take measures to prevent further cases. Certain tickborne diseases such as anaplasmosis and ehrlichiosis have not been known to be transmitted to humans within Washington. If an investigation found a case without a history of travel, alerts could be issued to warn the public of the potential for local transmission and risk activities, and to inform providers how to diagnose cases.

Finally, some conditions may be considered rare diseases of public health significance due to their potential use for intentional dissemination. Suspected person-to-person spread of vaccinia, the smallpox vaccine virus, are investigated to confirm that transmission was not intentional and that the virus is in fact vaccinia, not smallpox.

**Reported Rare Diseases**

Many diseases that are now more common or are well-understood were once unknown. Legionellosis was recognized in 1976 after an outbreak of infections associated with a hotel convention. The first cases of HIV infection were reported in 1980. Hantavirus infections with the North American Sin Nombre virus were first recognized in 1993 with an outbreak in the Four Corners area. In each situation, clinicians reported patients with unusual syndromes which prompted public health investigations. Initial reports led to the identification of the causative agents, associated illnesses, and risk exposures.

There is particular concern for conditions that have outbreak potential or that could become established in the region. Avian influenza is such a condition. There is ongoing international surveillance for new influenza strains that could cause a worldwide pandemic.

In contrast, highly antibiotic resistant organisms are unlikely to cause an outbreak, but cases may infect other patients in a healthcare setting, can be complicated to treat, and may result in death. Once carbapenam-resistant Enterbacteriaceae (CRE) were documented in Washington, voluntary surveillance was initiated. When a CRE case is laboratory confirmed, the healthcare facility undertakes infection control measures to prevent additional infections. Coccidioidomycosis and *Cryptococcus gattii*, two rare fungal diseases where a minority of Washington’s cases are locally exposed, are also voluntarily reported as rare diseases of public health significance.

**Carbapenem-Resistant Enterbacteriaceae Isolates, Washington 2015-2016**

![CRE Quarterly Report, December 2016](image)
In Washington, only a small number of rare infectious diseases are identified and reported to public health authorities. These reports permit our public health system to track the diseases with endemic exposures in Washington, to track travel-associated diseases, and to provide resources for providers and local health jurisdictions when cases occur.

Rare diseases of interest include cases that may or may not have been previously reported to a local public health agency and may be either locally acquired or travel-related:

- Reported, endemic to Washington: babesiosis, spotted fever rickettsiosis, tick paralysis
- Reported, travel-associated: African sleeping sickness, Chagas disease
- Never reported but of concern if identified: MERS, human cases of novel influenza

Note that a WAC revision is underway that plans to list the rare diseases currently in the definition (http://apps.leg.wa.gov/wac/default.aspx?cite=246-101-010) as distinct notifiable conditions (http://apps.leg.wa.gov/wac/default.aspx?cite=246-101-101). The general category of public health events of concern will remain notifiable with the goal of detecting newly emerging diseases.

The readiness of healthcare providers to notify public health agencies of unusual illnesses can benefit the patient and the general community. Correct treatment, prompt prevention measures, and public education may be initiated for a suspected or confirmed rare disease to protect the public’s health.

**Resources**


