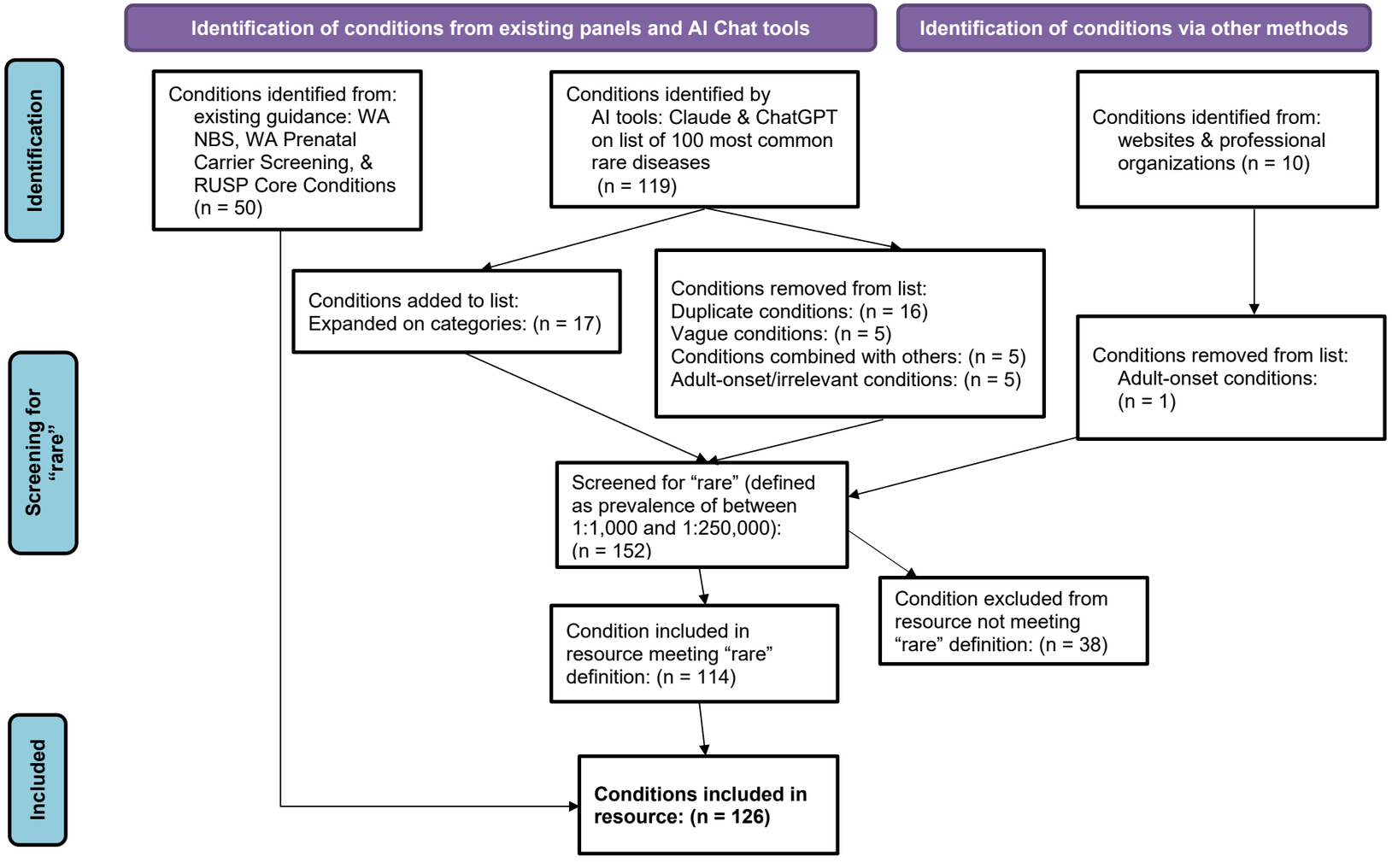
Rare Disease Resource – Inclusion and Exclusion Criteria
Genetics Program



From: Page MJ, McKenzie JE, Bossuyt PM, Boutron I, Hoffmann TC, Mulrow CD, et al. The PRISMA 2020 statement: an updated guideline for reporting systematic reviews. BMJ 2021;372:n71. doi: 10.1136/bmj.n71.

This document outlines the process we used to select the 126 rare diseases in the Rare Disease Resource created by the Washington State Department of Health (WA DOH). Below is an overview of our process:

- First, we used 2 AI tools to list the 100 most “common” rare diseases. We then added 10 more diseases identified through professional organizations and other sources.
- Next, we applied criteria to optimize our list. We focused on including more diseases by expanding on specific categories of conditions. We excluded diseases that were adult-onset, duplicates, combined conditions or did not meet the definition of rare as determined for this resource (prevalence of 1 in 1,000 to 1 in 250,000). This process gave us a list of 114 diseases.
- We then included conditions screened in Washington as part of the prenatal carrier screening, newborn screening, and those recommended by national guidelines from the Recommended Uniform Screening Panel as of June 2024. This step added 12 more diseases.
- Some of these conditions did not meet this resource’s criteria for rare, but they were included since WA DOH screens or recommends screening for them.
- The final resource includes information on 126 rare diseases.

Washington State Department of HEALTH

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