Public Health Genomics: Reaching the Summit

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Objectives

- Define genomics in the context of public health
- Describe examples of how genomics can be integrated into public health programs
- Discuss the role of public health in the emerging field of personalized medicine


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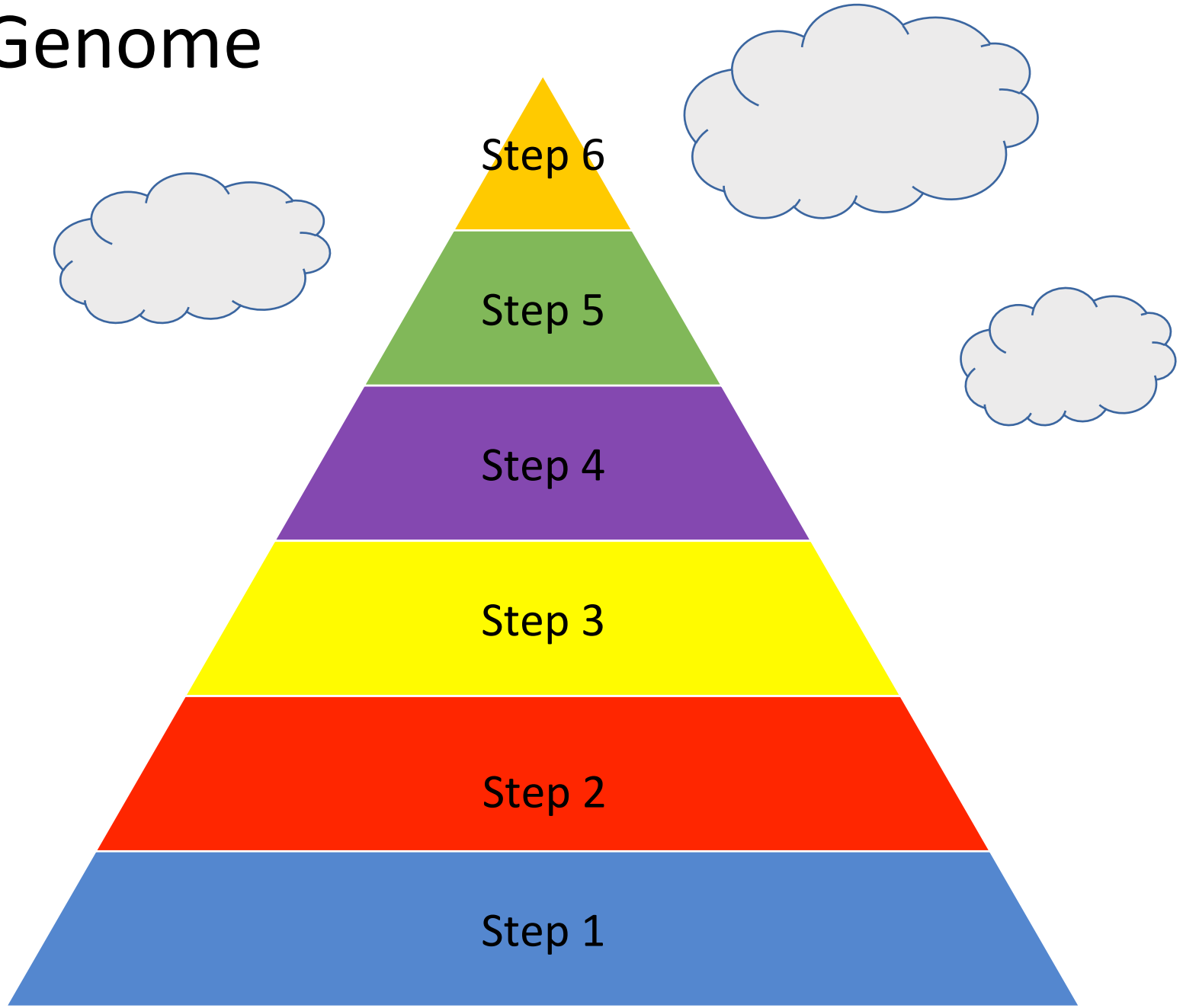
Mount Genome



Step 1: Chart our Path

What is our path? 10 essential PH functions:

- **Monitor** health status
- **Diagnose and investigate** health problems and hazards
- **Inform, educate and empower** people about health issues.
- **Mobilize** community partnerships to identify and solve health problems.
- **Develop policies and plans** that support individual/ community health efforts.
- **Enforce** laws and regulations that protect health and ensure safety.
- **Link** people to health services; assure provision of care when otherwise unavailable.
- **Assure** a competent public health workforce.
- **Evaluate** effectiveness, accessibility and quality of population-based health services.
- **Research** for new insights and innovative solutions to health problems.

(https://www.cdc.gov/publichealthgateway/publichealthservices/essentialhealthservices.html)

Step 2: Check our gear

PH Infrastructure, Tools and Resources

- Registries
  - Birth, death
  - Cancer
  - Other chronic disease
- Population-based surveys
  - BRFSS, PRAMS, provider access
- Prevention services
  - Cancer screening (breast, cervical, colon)
  - Wise Women program: CVD
  - Diabetes and CVD management
  - Clinical guidelines
- PH workforce training
  - Clinical Quality Improvement Program
  - Access to primary care providers, community clinics, hospitals, local PH
- PH Communications
  - Multi-media platforms
  - Public awareness campaigns
- Community Partnerships
  - Cancer coalition, local PH, community clinics, Medicaid/Medicare, hospitals
- Evaluation staff and expertise

Step 3: Comply with rules

Recommendations for Genomic Applications in PH

- 1997, CDC Office of Public Health Genomics (OPHG) established
- ***OPHG provides timely and credible information for the effective and responsible translation of genome-based discoveries into public health & health care***
- 2012, OPHG established system for evaluating 'readiness' of genomic applications based on available evidence (categorized into Tiers 1-3)
- 3 Tier 1 Applications = ready to go. **'Have significant potential for positive impact on public health based on available evidence-based guidelines and recommendations'**
  - FDA label requires use of test to inform choice or dose of a drug
  - FDA cleared or approved companion diagnostic device
  - CMS covers testing
  - Clinical practice guidelines based on systematic review supports testing

https://www.cdc.gov/genomics/implementation/toolkit/tier1.htm

Tier 1:Hereditary breast and ovarian cancer

- ***Women should be screened for family history that may be associated with an increased risk for potentially harmful mutations in breast cancer susceptibility genes (BRCA1 or BRCA2). Women with positive screening results should receive genetic counseling and, if indicated after counseling, BRCA testing.***

- 2005/13 USPSTF Guideline (https://www.uspreventiveservicestaskforce.org/)
- Healthy People 2020 Objective (https://www.healthypeople.gov/2020/topics-objectives/topic/genomics)

Tier 1: Lynch syndrome (LS)

- ***All people with new diagnosed colorectal cancer should be offered genetic testing for LS to reduce morbidity and mortality in relatives***.

- 2009, EGAPP recommendation (https://www.nature.com/articles/gim20095)
- Healthy People 2020 Objective (https://www.healthypeople.gov/2020/topics-objectives/topic/genomics)

Tier 1: Familial Hypercholesterolemia

- ***Cascade screening using cholesterol testing with or without DNA analysis should be conducted on relatives of affected persons with FH in order to identify previously unknown cases of FH and provide those people with life-saving treatment***

- NICE recommendation, 2013 (https://www.nice.org.uk/guidance/qs41)

Tier 1 Applications affect ~2 million Americans

- Population Health Impact:
  - **HBOC**: mutation prevalence = 1 in 300 to 1 in 500; account for 5-10% of breast, 15% ovarian cancers; 40-80% lifetime risk; increased risk for pancreatic, prostate cancer
  - **Lynch syndrome**: mutation prevalence = 1 in 300; 3-5% of colorectal cancer; up to 80% lifetime risk; increased risk for endometrial, small bowel, liver, ovarian, pancreatic
  - **FH**: mutation prevalence = 1 in 250 to 1 in 500; 5x risk of coronary heart disease; early heart disease and heart attack
- Most people at risk, **do not know it**
- Evidenced-based **interventions exist** to reduce risk and morbidity

Step 4: Forge the trail

Integrate Tier 1 Applications into PH Practice

- Background: In 2003; CDC OPHG began funding a few states to enhance implementation of Tier 1 applications into PH
  - Michigan, Oregon, Washington, Georgia, Utah, Connecticut, Colorado
  - 3 Strategies: Education, Policy and Systems Change, Surveillance
  - Focus on HBOC and more recently LS and FH
  - 2014 OPHG developed tool-kit for states to adopt these strategies

    https://www.cdc.gov/genomics/implementation/toolkit/tier1.htm

Colorado Experience: Getting started

Colorado Cancer Genetics Alliance
Reducing the Burden of Cancer in Colorado

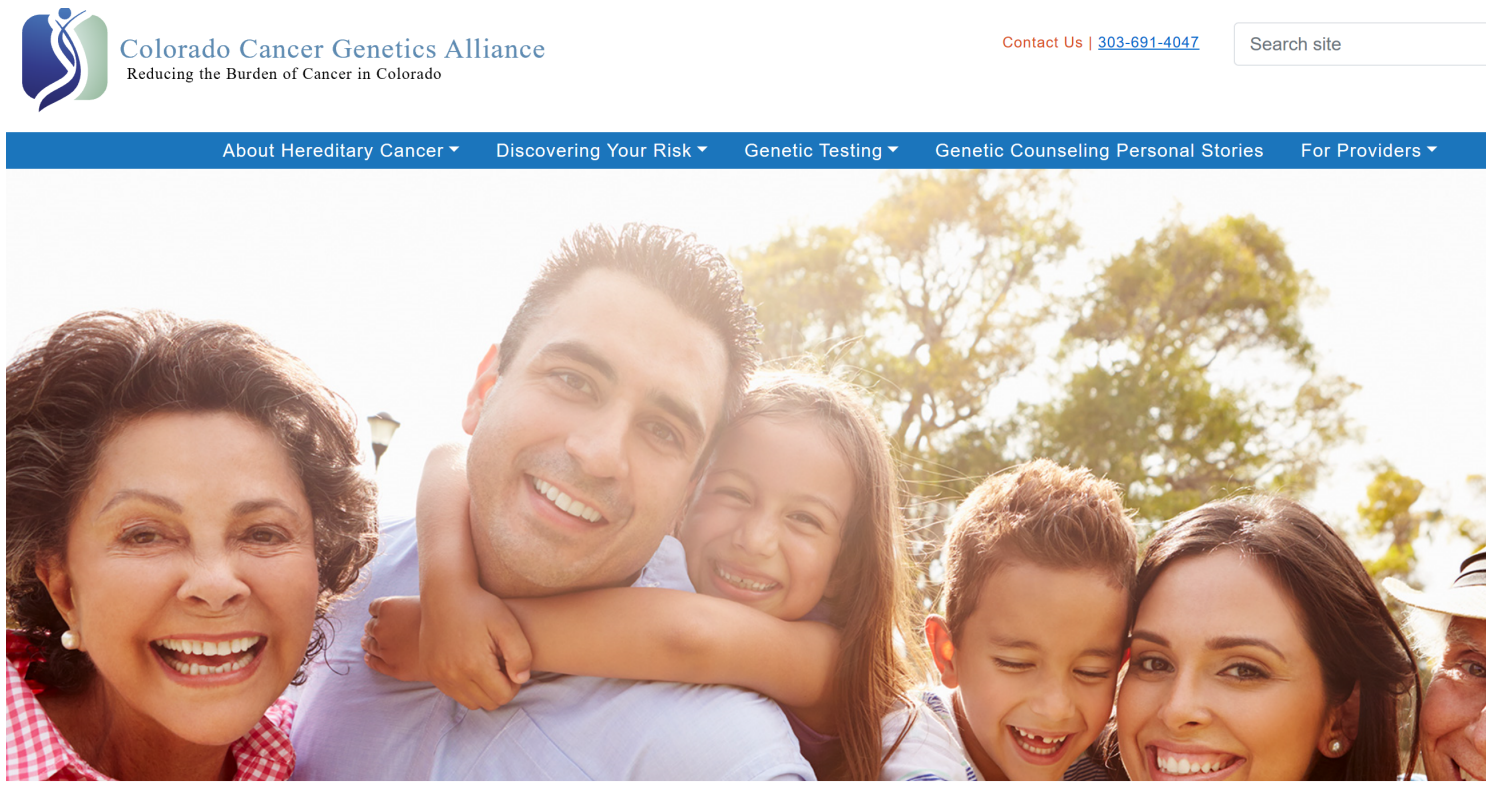
- Build infrastructure at CDPHE
  - Hire Genomics Coordinator
  - Establish shared staffing model with: cancer registry, comprehensive cancer, health informatics, communications, program evaluation

- Establish external partnerships
  - Univ Colorado Cancer Center
  - CO School of Public Health
  - Cancer Coalition/ Family History Task Force
  - Community Advisory Committee
  - Healthcare systems

Colorado Experience: Education

Goals:

- Increase public awareness
  - website, social media, video
- Increase provider awareness and knowledge about hereditary cancer
  - webinars, presentations to providers and professional societies
  - In-person training: Cancer Genetics: *Why It Matters for Primary Care Practice*



www.cocancergenetics.org

Gene Video

(https://www.youtube.com/watch?v=jN_jGoHmjZc&t=186s)

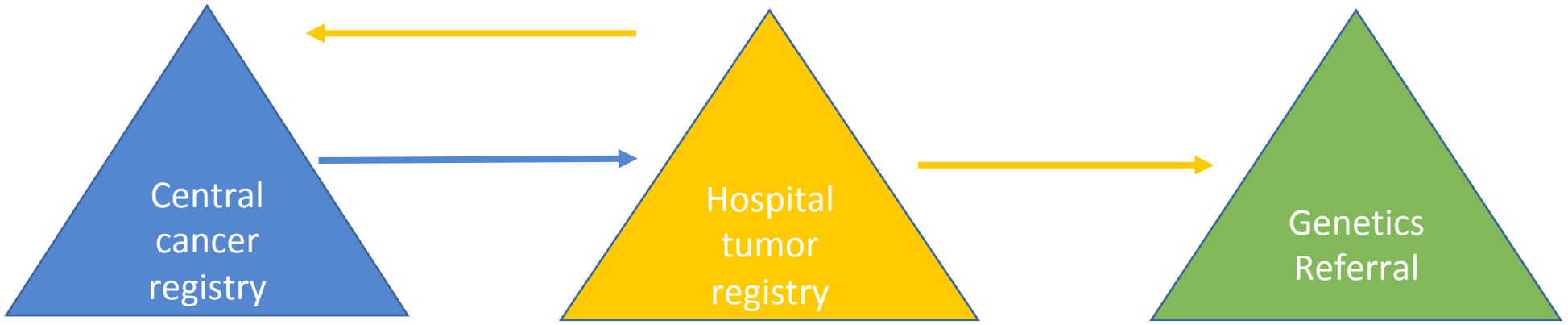
Colorado Experience: **Policy and Systems Change**

Colorado Cancer Genetics Alliance
Reducing the Burden of Cancer in Colorado

- Bi-directional reporting pilot
- Family history screening
- LS tumor testing

Bi-directional Reporting

- Goal= identify cancer survivors at risk for HBOC/LS and increase referrals for genetic services



Run algorithm to ID at-risk cancer survivors

Check patient EMR for referral/testing status
STOP: use for quality improvement purpose

Notify providers/ patients; refer for genetic services

Bi-directional Pilot Results

	Institution 1		Institution 2		Institution 3		All institutions	
	Total number cases	Number referred and/or tested	Total number cases	Number referred and/or tested	Total number cases	Number referred and/or tested	Total number cases	Number referred and/or tested
Breast cancer	42	28 (66.7%)	294	96 (32.5%)	308	282 (91.5%)	644	**406 (63%)**
Ovarian cancer	4	4 (100%)	55	23 (41.8%)	87	78 (89.7%)	146	**105 (72%)**
Colon cancer	*5	*4 (80%)	*65	*15 (23.8%)	91	65 (71.4%)	*168	***90 (54%)**
Uterine cancer					7	6 (85.7%)		

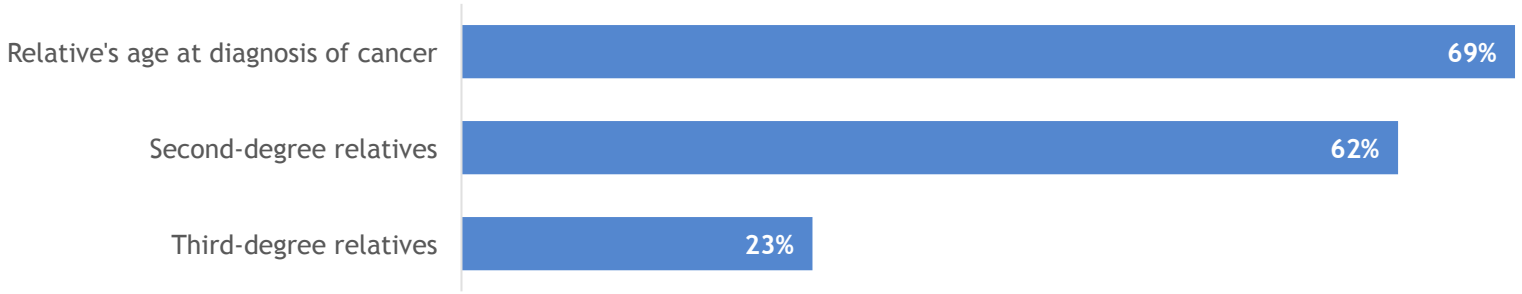
***Combined colon and uterine cancer**

Family History Screening

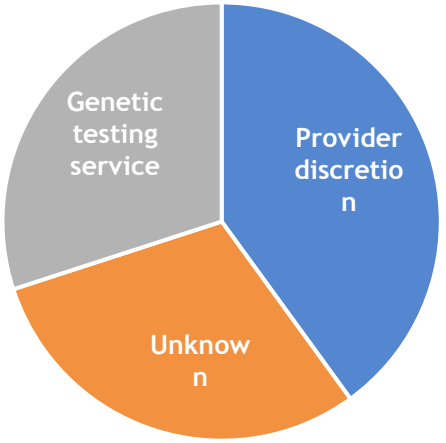
Colorado Cancer Genetics Alliance
Reducing the Burden of Cancer in Colorado

Goal = facilitate implementation of family history screening tool into primary care clinics

Survey of community clinics in CO to assess current practices:



Family history routinely collected



Referral practices for genetics

**Most clinics do not collect adequate family history to refer patients for screening or genetics

LS Tumor Testing

Colorado Cancer Genetics Alliance
Reducing the Burden of Cancer in Colorado

- Surveyed 44 Colorado hospitals to assess current practices
  - 79% reported that they screen all colon cancers
  - 54% have a written policy for universal Lynch screening (ULS)
  - Fewer rural hospitals are screening

Next steps:

- Develop information about ULS implementation for hospitals
- Develop report cards for hospitals using data from central cancer registry data (MSI, IHC for colon and endometrial tumors)

Colorado Experience: Surveillance

Colorado Cancer Genetics Alliance
Reducing the Burden of Cancer in Colorado

- **Hereditary cancer burden**: Central cancer registry
  - Algorithm to identify survivors at increased risk for HBPC/LS based on guidelines
  - New** added fields for 'referred for genetic counseling and/or testing' to abstract

- **Utilization of genetic services**: All Payor Claims data

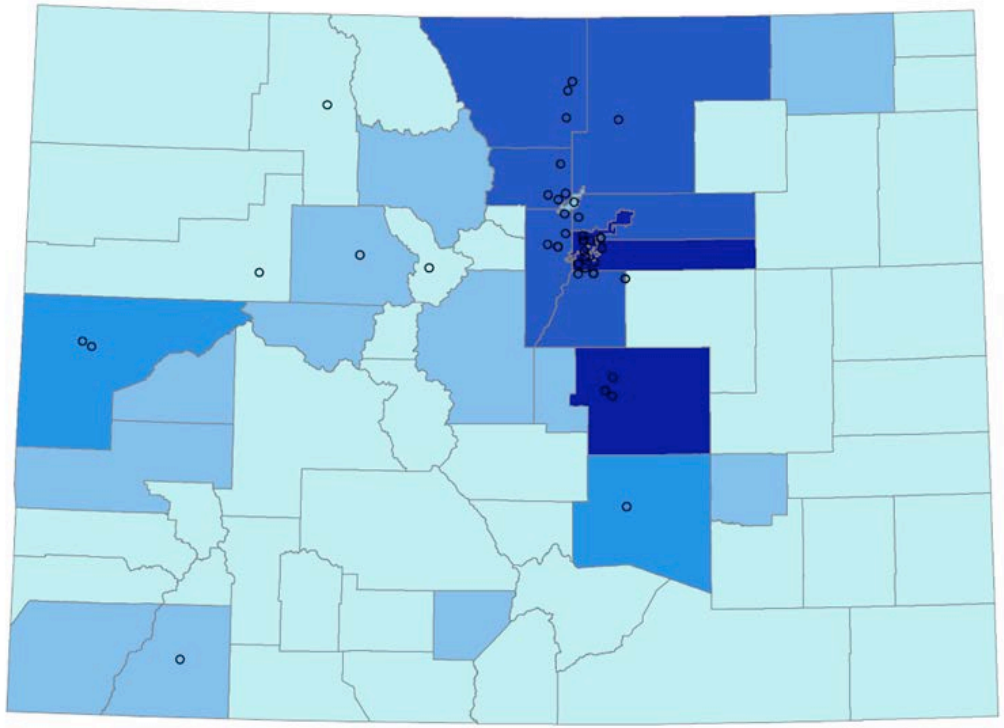
- **Prevalence of cancer family history and referral to genetics; family history communication; screening compliance among high risk**:
  - BRFSS and PRAMs surveys
  - Added 6 questions to BRFSS, 2016 and 2018

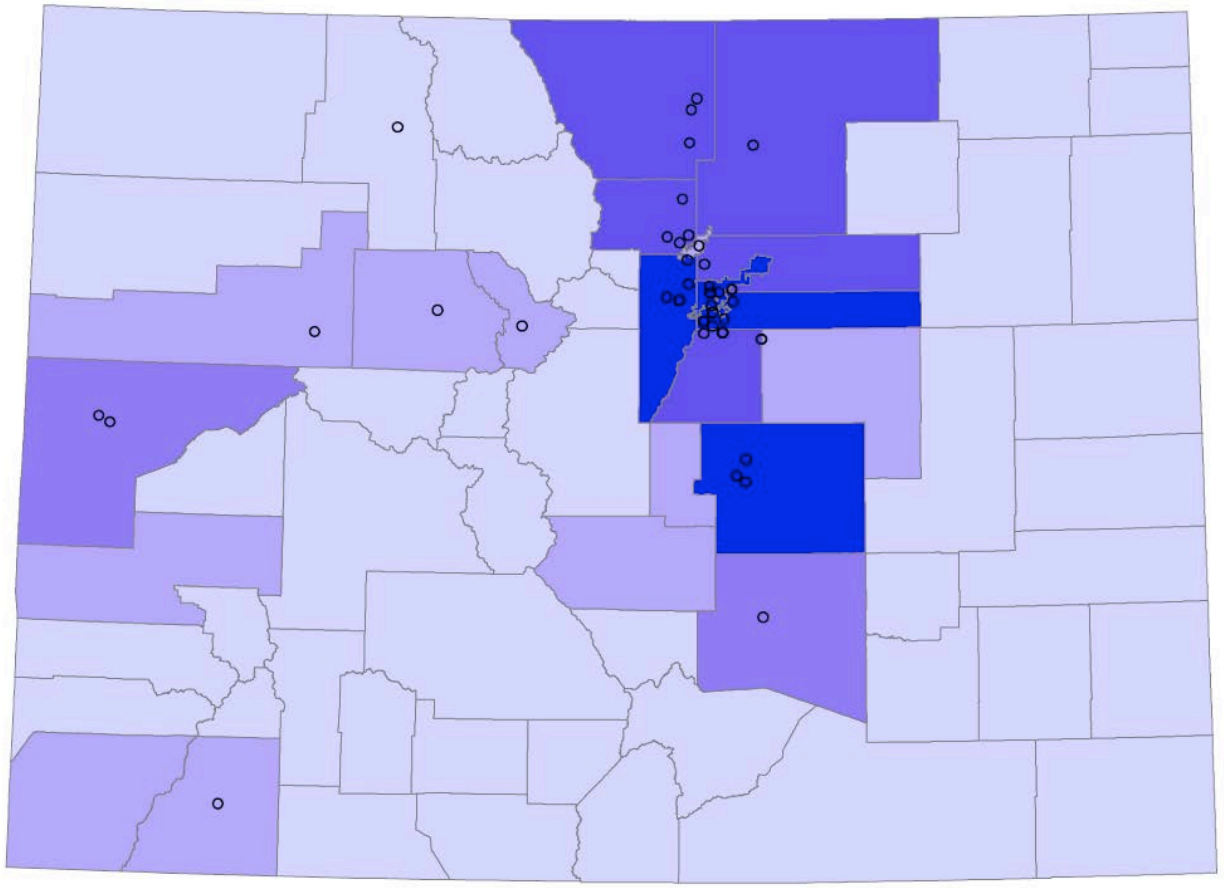
Hereditary Cancer Burden in Colorado (per NCCN guidelines)

Syndrome / Specific Criteria	People meeting criteria		People meeting criteria 'exclusively'	
	N	%	N	%
HBOC				
Breast cancer <=50	32375	45.8%	27022	38.2%
Two breast cancer primaries	8704	12.3%	5320	7.5%
Breast cancer <=60 that is triple negative for ER/PR/Her2	720	1.0%	241	0.3%
Male breast cancer	662	0.9%	543	0.8%
Ovarian cancer at any age (epithelial, non-mucinous)	13303	18.8%	12267	17.4%
Metastatic prostate cancer	7692	10.9%	7666	10.8%
Ashkenazi Jewish decent with breast, ovarian or pancreatic cancer at any age	319	0.5%	174	0.2%
Breast and pancreas any age	407	0.6%	263	0.4%
Lynch Syndrome				
CRC <50	6177	8.7%	5536	7.8%
CRC at any age that is MSI unstable or MMR gene deficient	514	0.7%	358	0.5%
Endometrial <50	2580	3.7%	2318	3.3%
Endometrial ca at any age that is MSI unstable or MMR gene deficient	NA yet		NA yet	
CRC with metachronous or synchronous LS cancer*	1987	2.8%	802	1.1%
Endometrial ca with metachronous or synchronous LS cancer*	1216	1.7%	152	0.2%
Synchronous/Metachronous CRC	2697	3.8%	2017	2.9%

Availability of cancer genetics counselors in CO

Distribution of cancer cases: Breast cancer <50, ovarian cancer

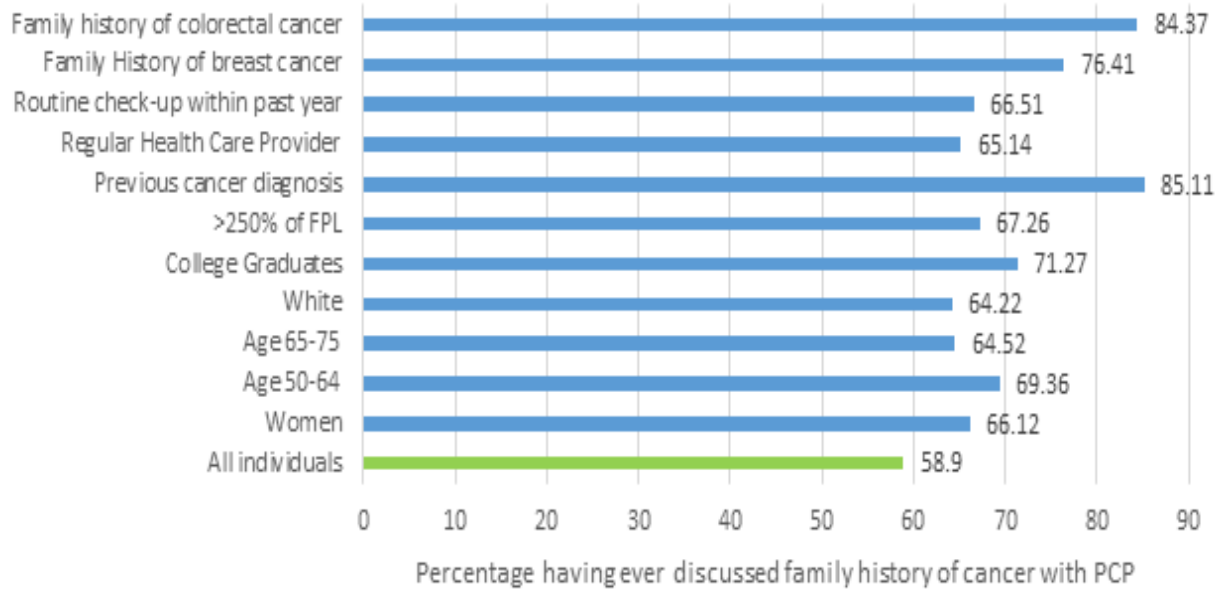


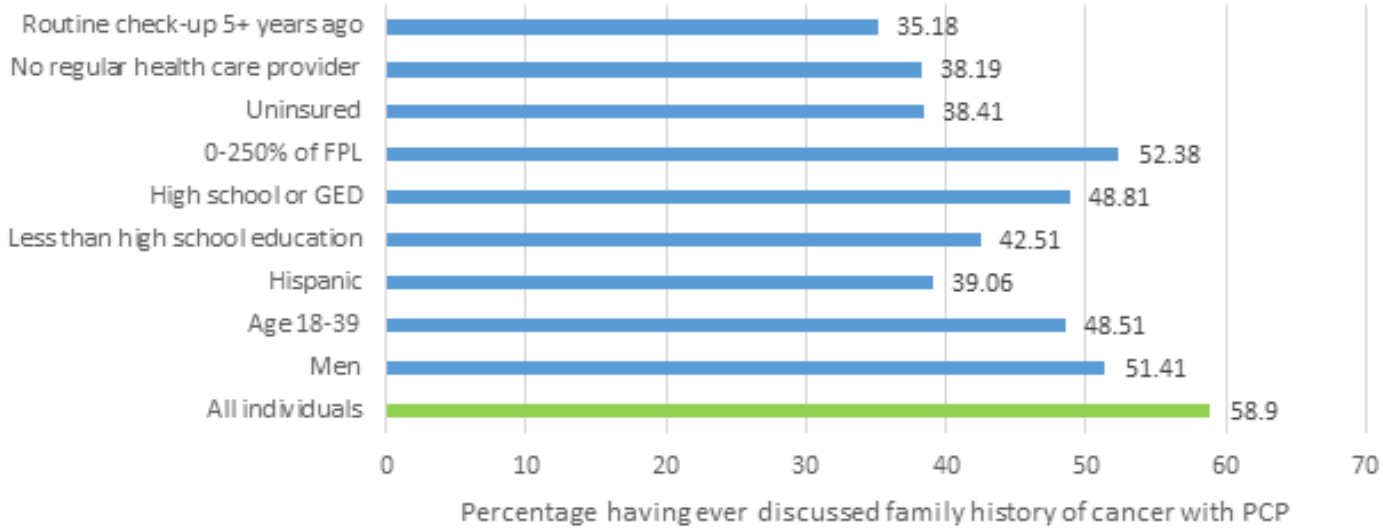


Distribution of cancer cases: colon <50, uterine cancer <50

BRFSS Results

- Fam Hx breast/ovarian ca <50: 11%
- Fam Hx colon ca <60: 6%
- If YES, referred for genetics: ~60%

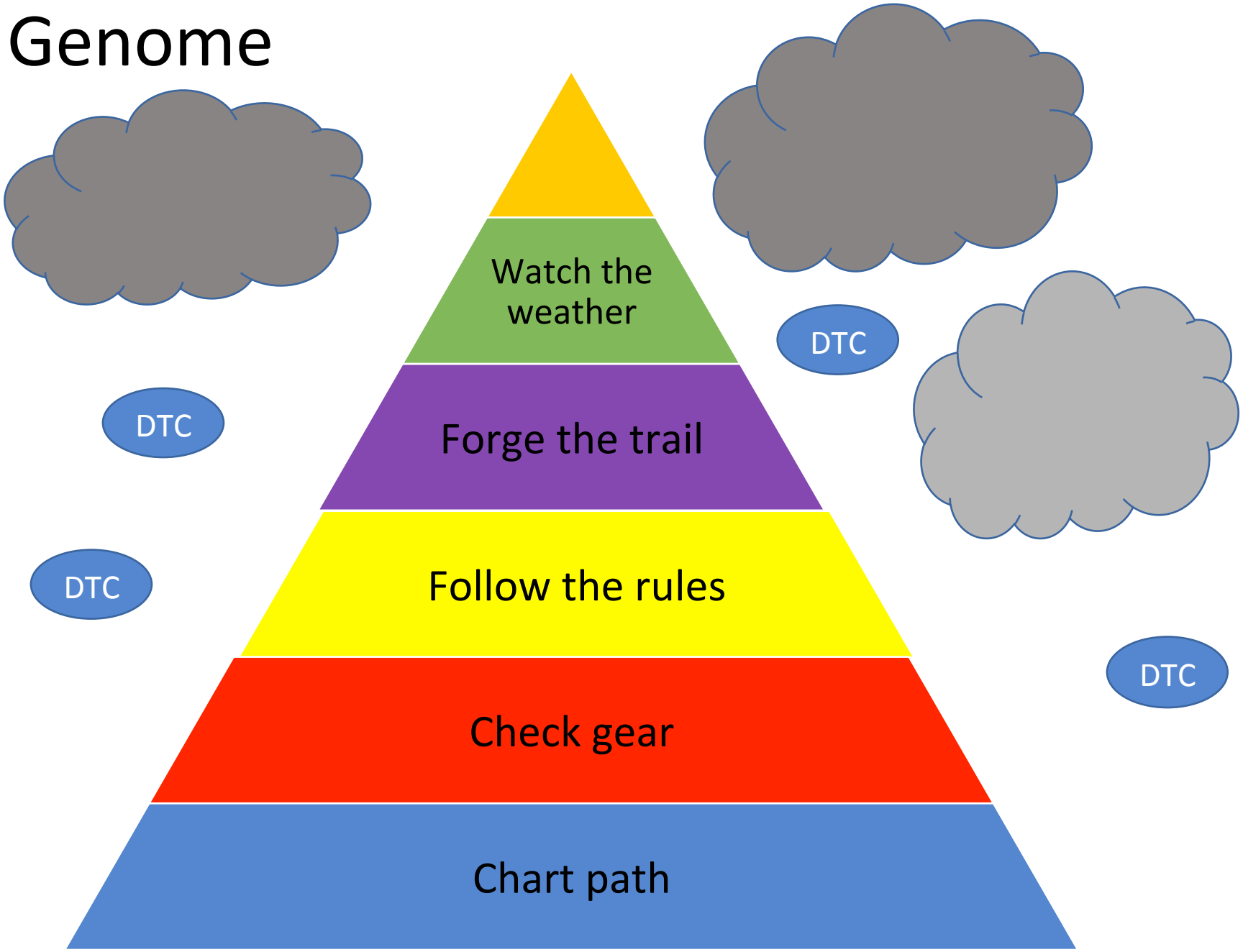




Have you ever spoken with your medical provider about your family history of cancer?

Step 5: watch the weather

Mount Genome



Changing Climate: emergence of DTC genetic tests and personalized medicine

- >12 million people in US have used DTCs and #s are growing
  - Precision Medicine Initiative – *All of Us*
  - Biobanks
  - Growing disparities in awareness and knowledge (Am J Prev Med 2018;54:6:806-13)

- What is the role of PH?
  - **Inform, educate and empower**
    - Interpreting DTC test results – 'no' news is not necessarily 'good' news
    - Provide perspective: weight of genomics vs other risk factors, e.g. obesity
  - **Link** people to services; assure provision of care when otherwise unavailable
    - Establish central resource for genetics providers accessible to all
  - **Assure** a competent public health workforce
    - Need for improved genomic literacy among PH workers, providers, students
  - PH must be involved!!

Step 6: Finish strong

Sustainability: how do we assure that genomics stays integrated into PH practice?

- Problem:
  - Outside of OPHG grants, no direct money for ‘genomics’ for state PH
  - Majority of PH funding is federal (CDC); small % is state monies

- Solutions?
  - Specific tax, like tobacco tax in CO. What would be taxed?
  - Extend partnerships with non-for-profit groups, e.g. Foundations
  - Establish public-private partnerships, e.g. with testing laboratories?
  - Re-direct existing funding (from state and CDC) to integrate genomics across multiple PH programs; e.g. cancer registry, comp cancer, cancer screening programs, chronic disease and wellness programs

Reaching the Summit

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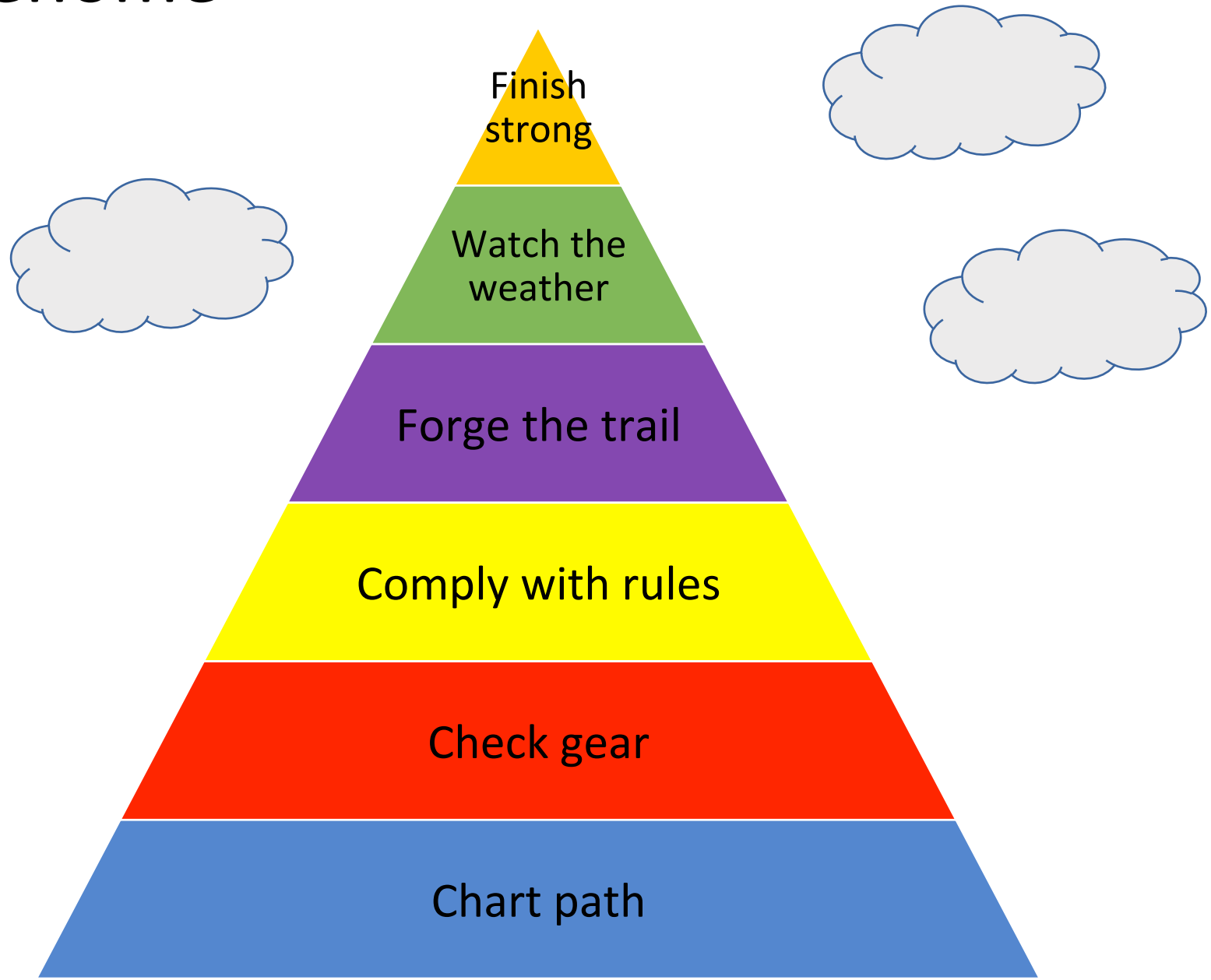

Bike lanes were made to provide a boundary between bikers, pedestrians and vehicles. If rules are followed this can help keep people out of harm's way.

Construction sites with signs posted can alert the community of a potential danger zones and to proceed with caution.

photo by Gabrielle M

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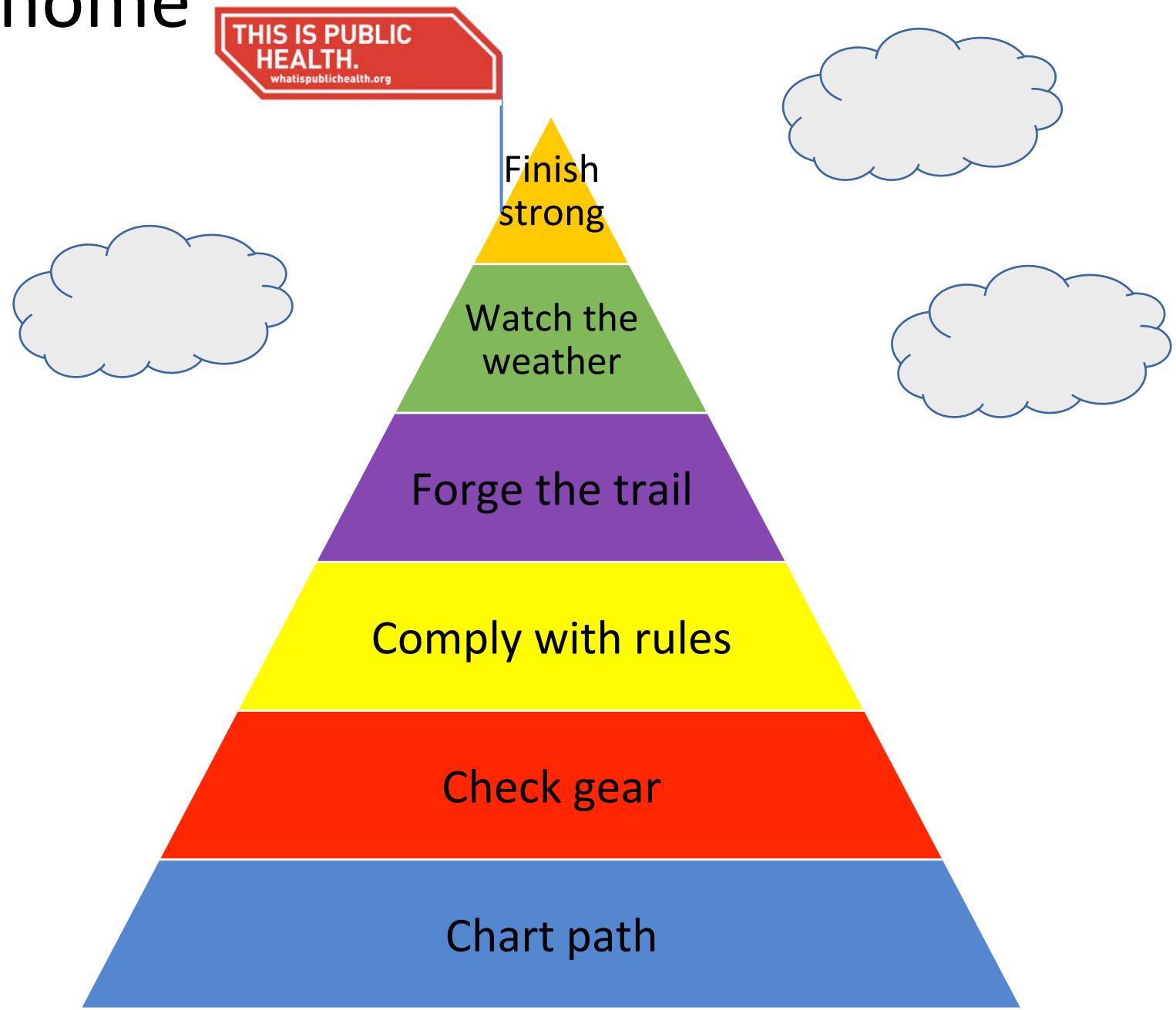

Acknowledgements

CCGA Team

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