# Development of a standard of care for patients with valosin-containing protein (VCP) associated multisystem proteinopathy (MSP)

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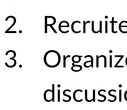

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**AIM 1:** Establishing a multidisciplinary standard of care for appropriate pharmacotherapies and supportive therapies
**AIM 2:** Expediting time to accurate diagnosis
**AIM 3:** Identify gaps and future directions for clinical research

## METHODS

1. Recruited a multidisciplinary team of 50 physicians and therapists
2. Domain teams reviewed literature, exchanged ideas, and prepared a domain consensus recommendation based on expert opinion and adjacent disease practices
3. A virtual consortium meeting was held on April 9, 2021
4. Meeting discussion points integrating into one manuscript with team member sign-off

## PATIENT ADVOCACY ROLE

1. Provided patient perspective in project scope
2. Recruited expert clinicians to participate
3. Organized communications, facilitated discussions, and hosted meetings
4. Assisted in literature review
5. Reviewed and edited the manuscript concerning patient perspective and symptoms

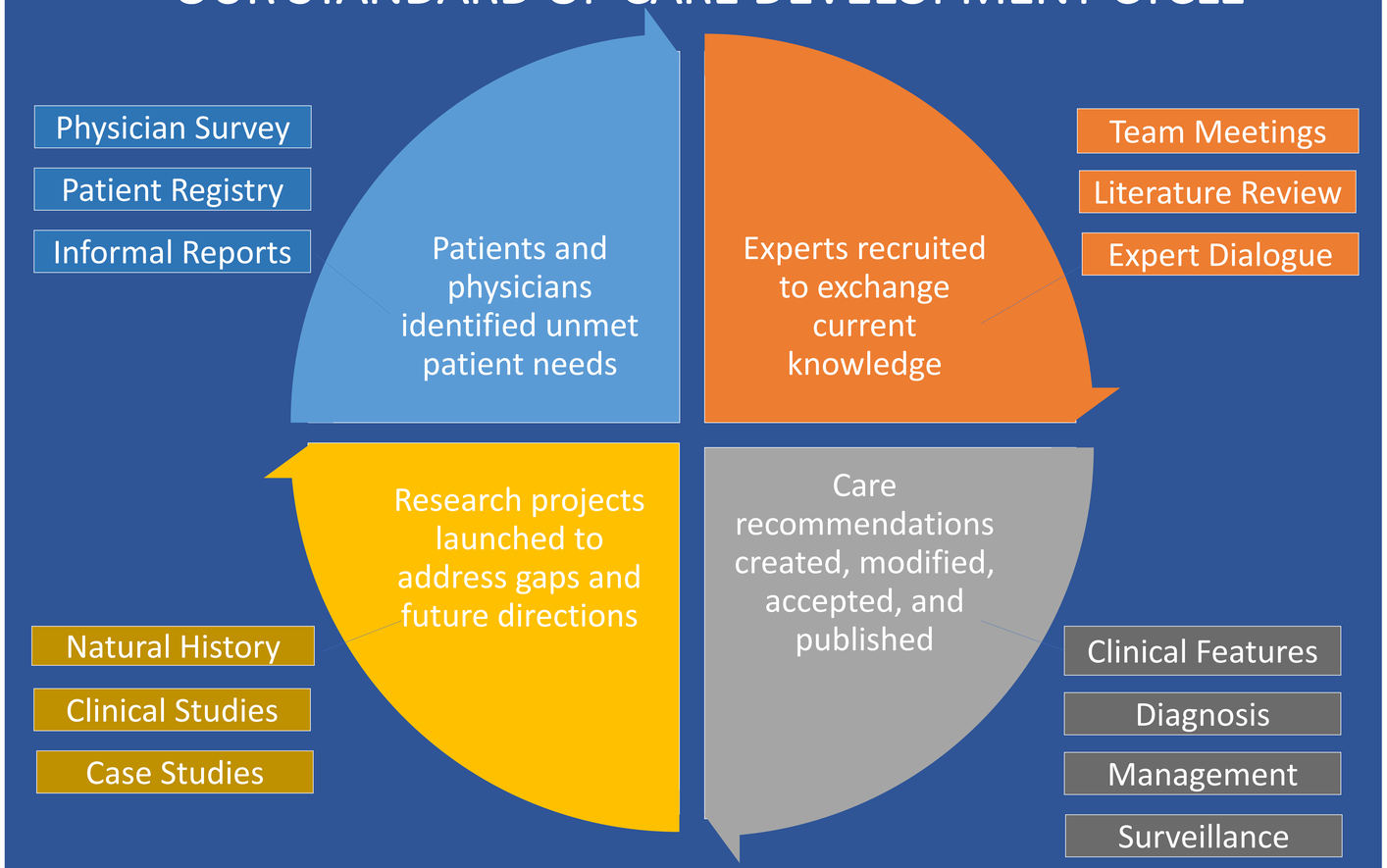
## RESULTS

- Each domain team created a 2-5 page consensus guideline
- One multidisciplinary manuscript has been submitted for publication

# International **collaboration** among a **multidisciplinary team** addresses **unmet patient need** in **rare disease:**

1. Delays in diagnosis and prolonged time to treatment
2. Delays in recognizing involvement of other organ systems
3. Disparate care between clinics
4. Disease development in at risk, undiagnosed family members

## OUR STANDARD OF CARE DEVELOPMENT CYCLE

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## ABOUT VCP ASSOCIATED MSP

Rare, heterogeneous, autosomal dominant, genetic disorder affecting multiple organ systems including the muscular, skeletal, and central nervous system

## PREVALENCE OF PHENOTYPES

- Inclusion Body Myopathy ~ 90%
- Paget's disease of Bone ~ 40%
- Frontotemporal dementia ~ 30%
- Respiratory dysfunction ~ 40-50%
- Amyotrophic lateral sclerosis ~10%
- Parkinson disease ~ 4%
- Alzheimer disease ~ 2%
- Spastic paraplegia ~ isolated
- Charcot Marie Tooth disease ~ isolated
- Cardiomyopathy ~ unknown
- Urinary and anal dysfunction ~ unknown

## MULTIDISCIPLINARY DOMAIN TEAMS

- Genetic diagnosis
- Myopathy
- Frontotemporal dementia
- Paget's disease of bone
- ALS and CMT
- Parkinson's disease/ parkinsonism
- Cardiomyopathy
- Respiratory dysfunction
- Supportive therapies [including physical and occupational therapy, speech language pathology]
- Mental health
- Supplements and nutrition

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