PATIENT REGISTRIES IN RARE DISEASES

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Objectives

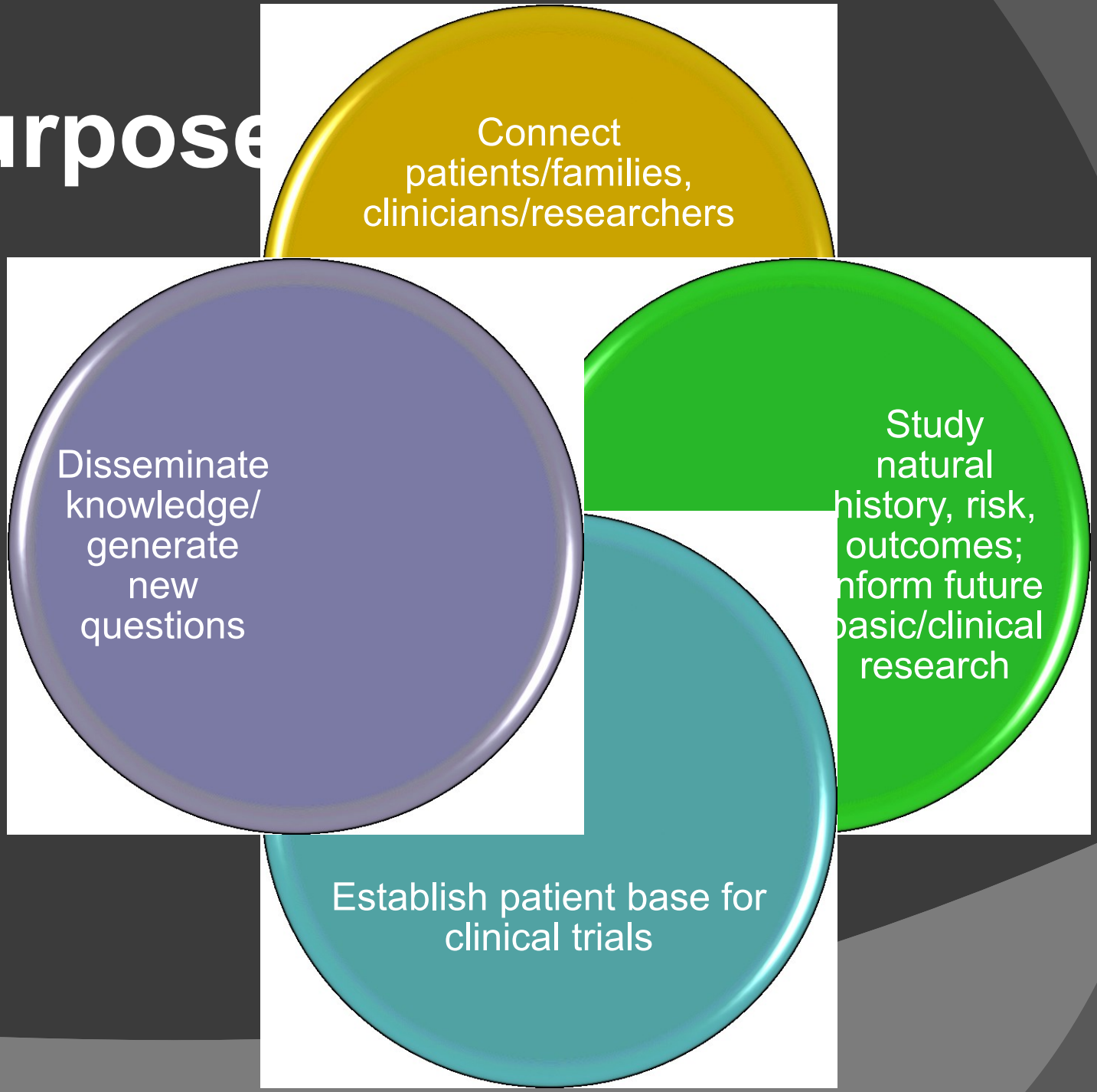
- Provide general overview and unique challenges of rare disease patient registries
- Discuss University of Rochester Batten Center (URBC) “registry” experience
- Inspire further enhancement and collaboration

Patient Registry Definition

An organized system that uses observational study methods to collect uniform data (clinical and other) to evaluate specified outcomes for a population defined by a particular disease, condition, or exposure; and serves one or more predetermined scientific, clinical, or policy purposes.

Gliklich RE, Dreyer NA: Registries for Evaluating Patient Registries: A User's Guide: AHRQ publication No. 07-EHC001. Rockville, MD. April 2007

Purpose


Connect patients/families, clinicians/researchers
Study natural history, risk, outcomes; inform future basic/clinical research
Establish patient base for clinical trials
Disseminate knowledge/ generate new questions

Registry Classification: Examples

- Disease-specific registries
  - Acute: MI, stroke
  - Chronic: asthma
  - Rare: CF, HD, JNCL, Hemophilia
  - Over a period of time: infection
- Clinical (e.g., encounter, procedure)
  - Outcomes
  - Safety surveillance

Registry Classification: Examples

- Humanistic & economic outcomes
  - Patient-reported outcomes
  - Compliance
  - Cost effectiveness
- Product
  - Post-approval/marketing product safety assessment

Registry Content/Organization

Varies from simple to vast network ("repository of registries")

- Contact registry
- Clinical data
  - History (Medical Records)
  - Exam
  - Laboratory tests
- Biospecimen repository
- Imaging
- Patient-supplied data
  - Surveys
  - Longitudinal outcomes
- Clinical Trial Databases
- Pharmacology
  - PD, Pk
- Patient Genomic data

"We don't know the questions that are going to be asked tomorrow," but without today's data, those questions could not be asked nor answered."

--Benjamin M. Greenberg, MD

Registries *vs.* Randomized Clinical Trials

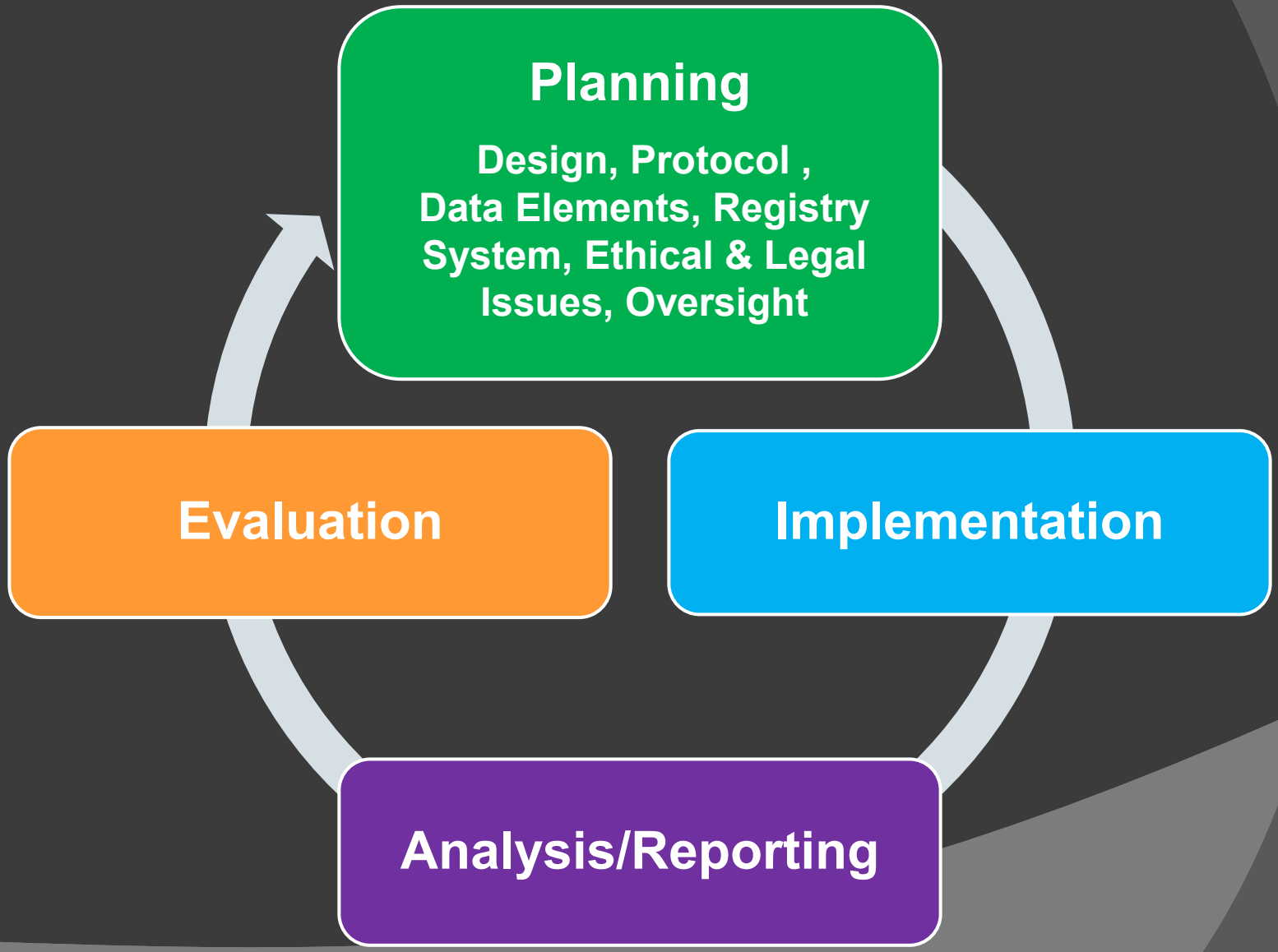
Registry

- Flexible
- Larger “N”
- Effectiveness
- Observational/“Real world”
- Hypothesis generating
- Good Clinical Practice optimal

RCT

- Specific
- Smaller “N”
- Efficacy
- Randomized/controlled
- Hypothesis driven
- Good Clinical Practice required

Registry Life Cycle: Planning



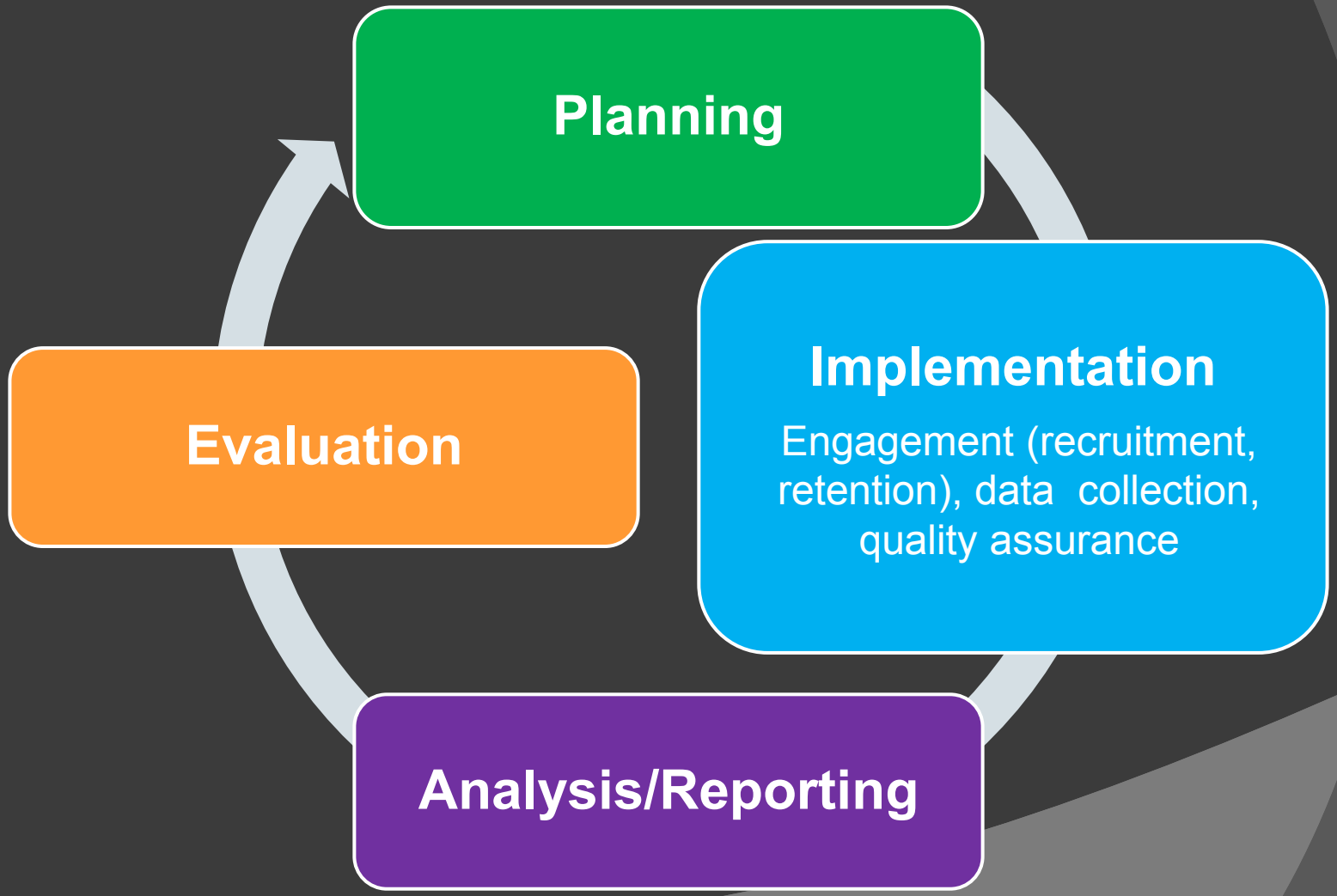
Planning Challenges

- Purpose, scope
- Stakeholders
- Infrastructure
- Funding
- Landscape
- Transparency

Data Management Planning

- Oversight (startup and maintenance)
- Data acquisition (primary, secondary sources)
- Data management system requirements
- Determine coding system for drugs, devices and/or medical events; dictionaries, if applicable
- Quality assurance at all levels

Registry Life Cycle: Implementation



Registry Life Cycle: Analysis/Reporting

Analysis & Reporting

- **Communication Plan**
  - Dissemination of results (investigators, participants, community)
    - Abstracts, papers
    - Journal authorship
    - Scientific/lay group meetings
  - Communicating progress, developments
    - Direct
    - Media
    - Web

Registry Life Cycle: Evaluation

URBC Registry to Clinical Trial

- UR IRB approved participant contact database
  - Any NCL
  - Parent consent for future contact
- Clinical Rating Scale (UBDRS)
  - Enabled natural history database
  - Concurrent data collection (medical history, medications, demographics)
  - Genotyping for NCL type, mutation
- Neuropsychological studies
- Several lines of inquiry
- Clinical trial launch

Emerging Trends with Registry Data

- Combining with other data sources or registries
  - Electronic health records
  - Common identifiers
  - Common data elements
- Linking patient information without use of full identifiers
- Analysis of linked registry data sets

Summary

- Patient registries
  - Useful tool, multiple purposes
- Registry science is evolving
  - Good ‘registry’ practices guidance

References

- AHRQ Registries for Evaluating Patient Outcomes: A User's Guide, 2nd edition (September 2010) http://www.effectivehealthcare.ahrq.gov/ehc/products/420/1337/RegistriesforEvaluatingPatientOutcomes3rdEd_DraftReport_20121128.pdf

(3rd edition draft release for public comment--September 2012)

- Rare Disease Task Force Report on patient registries in the field of rare diseases: Overview of the issues surrounding the establishment, management, governance and financing of academic registries (June 2011)

http://ec.europa.eu/health/rare_diseases/docs/patient_registries_rev2011.pdf