

An Open Call for Unity in Rare Disease Awareness

Rare Disease Day 2017 – NIH, Bethesda MD

Sharing the RARE Puzzle

When working on your disease's rare puzzle you will find that some pieces are shared with across the rare disease community.

Two key shared parts of the puzzle are awareness and policy. Policy is the subject of a different poster and white paper.

For shared parts of the puzzle we must become disease agnostic linking our pieces to pieces in thousands of other puzzles. The mosaic of these pieces is intricate but the impact of the bigger picture dominates that of a single disease.



the rare community needs, and to help pay for therapies through funding basic science, research towards therapies, approval, and reimbursement. They see a disorganized or uninformed community and may use this to shy away from supporting us. The general public will never understand the subtleties behind the differences in these numbers.

The effectiveness of our rare disease awareness and support depends on united messaging.

RARE Disease Facts

- **7,000+** rare diseases
- **1 in 10** affected ... **over 30 million Americans**
- **over 50%** of which are children, **30%** of which will not live to their 5th birthday
- **<5%** of rare diseases have approved therapies

Unfortunately, as you can see in the table below, some of us are placing unmatched puzzle pieces by stating different numbers. This distracts and confuses the viewers of our puzzles ... policy makers and the general public who we need to better understand

A Call for Rare Fact Unity

MLD Foundation is calling on the rare disease community –public, private, corporate, and institutional – to sing harmoniously from the same song sheet.

We call for all to all quote the same simple facts (see Rare Disease Facts in 1st column) when doing general Awareness and basic fact descriptions of Rare Disease. We must sing from the same song sheet to maximize our impact when communicating with the general public, legislators, and the media.

We acknowledge there are times when precise scientific accuracy is required. If you are counting records in your database then be precise. But we will also argue there is no precise definitive rare disease count because more are being discovered all of the time and, as the table below reflects, there are country by country variations in how rare diseases are defined (see table below).

| Organization | Rare Count | Rare Percent |
|-----------------|---------------------|--------------------|
| Orphanet | 6,547 | |
| GARD (NIH/ORDR) | 6,000-7,000 (6,511) | 7-10% of Americans |
| NORD | 7,000 | 10% (1 in 10) |
| Global Genes | 7,000 (6,538) | 10% (1 in 10) |
| EuroDIS | 6,000-8,000 | 6-8% of Europeans |
| iRDiRC | 5,000-8,000 | 6-8% of Europeans |
| Advocacy | 7,000+ | 10% (1 in 10) |

| Country | Rare Disease Prevalence Defined as |
|---------------|------------------------------------|
| United States | 1 in 1,600 |
| Europe | 1 in 2,000 |
| Japan | 1 in 2,500 |

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1 Orphanet 2016 List of rare diseases and synonyms http://www.orpha.net/orphacom/cahiers/docs/GB/List_of_rare_diseases_in_alphabetical_order.pdf

2 GARD/ORDR/NCATS/NIH database counts 6511 diseases but qualifies not all of them are rare. Usually NIH states either 6,000 or 6,000-7,000 in presentations and publications. <https://rarediseases.info.nih.gov/diseases>

3 NORD - National Organization for Rare Disorders 2014 Rare Disease Fact Sheet http://cdn.rarediseases.org/wp-content/uploads/2014/11/NRD-1008-FactSheet_5.pdf and <https://rarediseases.org/for-patients-and-families/information-resources/rare-disease-information/>

4 Global Genes <https://globalgenes.org/rare-diseases-facts-statistics/> and <https://globalgenes.org/rarelist/>

5 EURODIS Rare Disease Fact Sheet http://www.eurordis.org/sites/default/files/publications/Fact_Sheet_RD.pdf

6 iRDiRC International Rare Diseases Research Consortium <http://www.eucerd.eu/upload/file/Reports/2014ReportStateofArtRDActivities.pdf>

7 Rare Disease Act of 2002