



Impact of Rare Diseases on Patients and Caregivers

A Patient/Caregiver Study by Rare Patient Voice

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Market Research Survey among Rare Patient Voice patient/caregiver panel

- N=1001
 - 408 patients
 - 511 caregivers
 - 82 both patients and caregivers
- Conducted Sept. 13-20, 2014
- US only

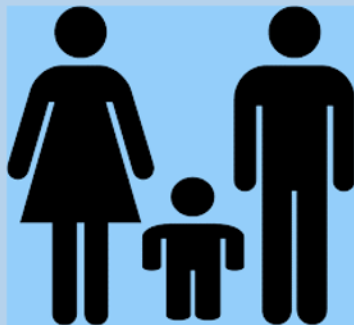


50+ Diseases Represented

22q11.2 deletion syndrome	Celiac disease	Hashimoto's Disease	Mastocytosis	Scleroderma
Afibrinogenemia	Common Variable Immune Deficiency (CVID)	Hemophilia	Meniere's	Sheehan's Syndrome
Agenesis of the Corpus Callosum	Crohn's	Hughes Syndrome.	Mitochondrial Disease	Sickle cell
Ankylosing Spondylitis	Cushing's Disease	Huntingdon's Disease	Narcolepsy	Spinal Muscular Atrophy
Aplastic Anemia	Cystic Fibrosis	Hydrocephalus	Osteogenesis Imperfecta	Sturge-Weber Syndrome
Arnold-Chiari Malformation Type 1 with Syringomyelia	Ehlers Danlos Syndrome	Hypoplastic Left Heart Syndrome	Pitt Hopkins syndrome	Systemic mastocytosis
Arthrogryposis	Factor VII Deficiency	Idiopathic Intracranial Hypertension	Pulmonary Arterial Hypertension	Ulcerative colitis
Autoimmune neutropenia	Gaucher Disease	Klippel Feil Syndrome	Recessive Dystrophic Epidermolysis Bullosa	Von Willebrand
Barth Syndrome	Glutaric Acidemia type 1	Lennox Gastaut Syndrome	Rett Syndrome	Waldenstrom Macroglobulinemia
CDKL5	Graves Disease	Marfan Syndrome	Schizencephaly	



Topics Covered: Rare Disease Impact



Impact on Family



Emotional Impact



**Financial/Employment/
Educational Impact**



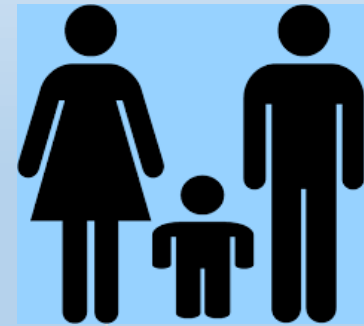
Impact on Social Interaction



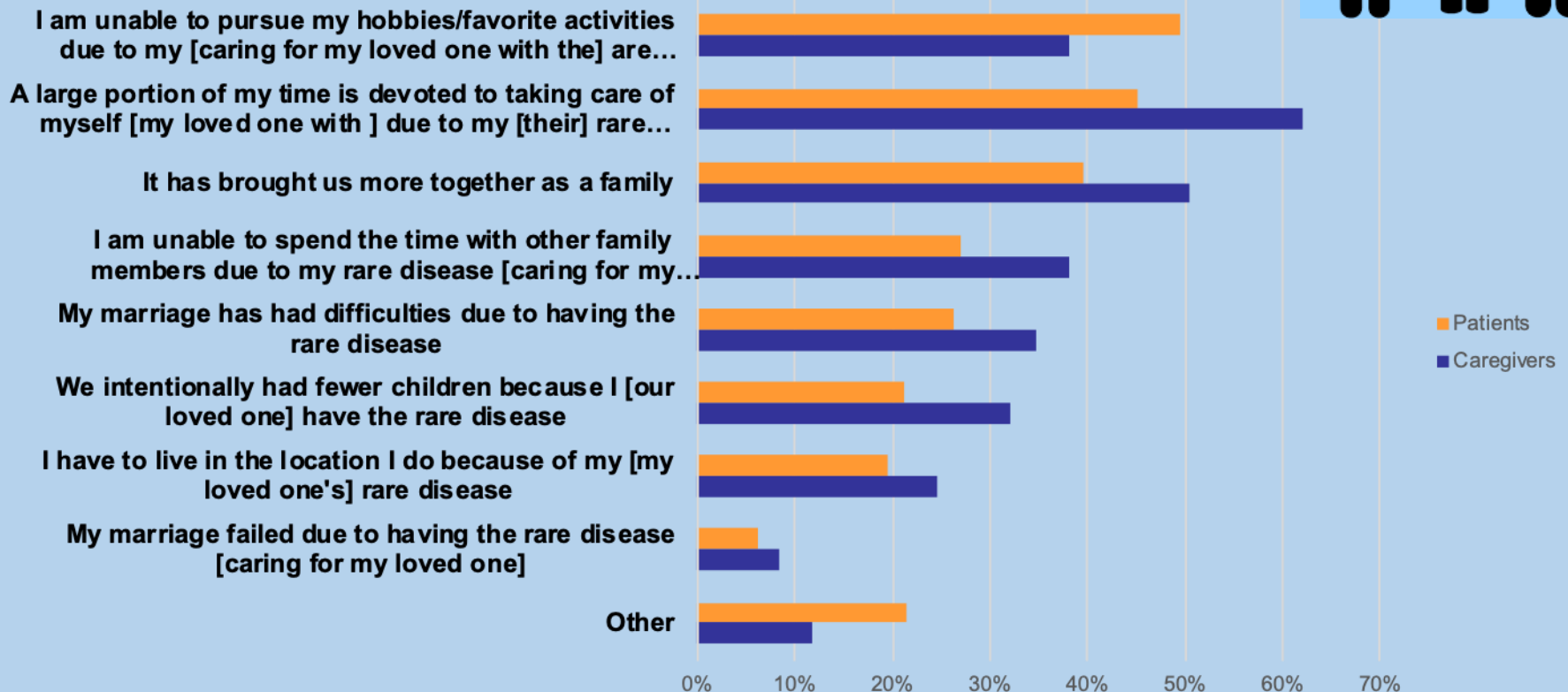
Effect of ACA/Obamacare



The impact on the family is especially hard on caregivers.



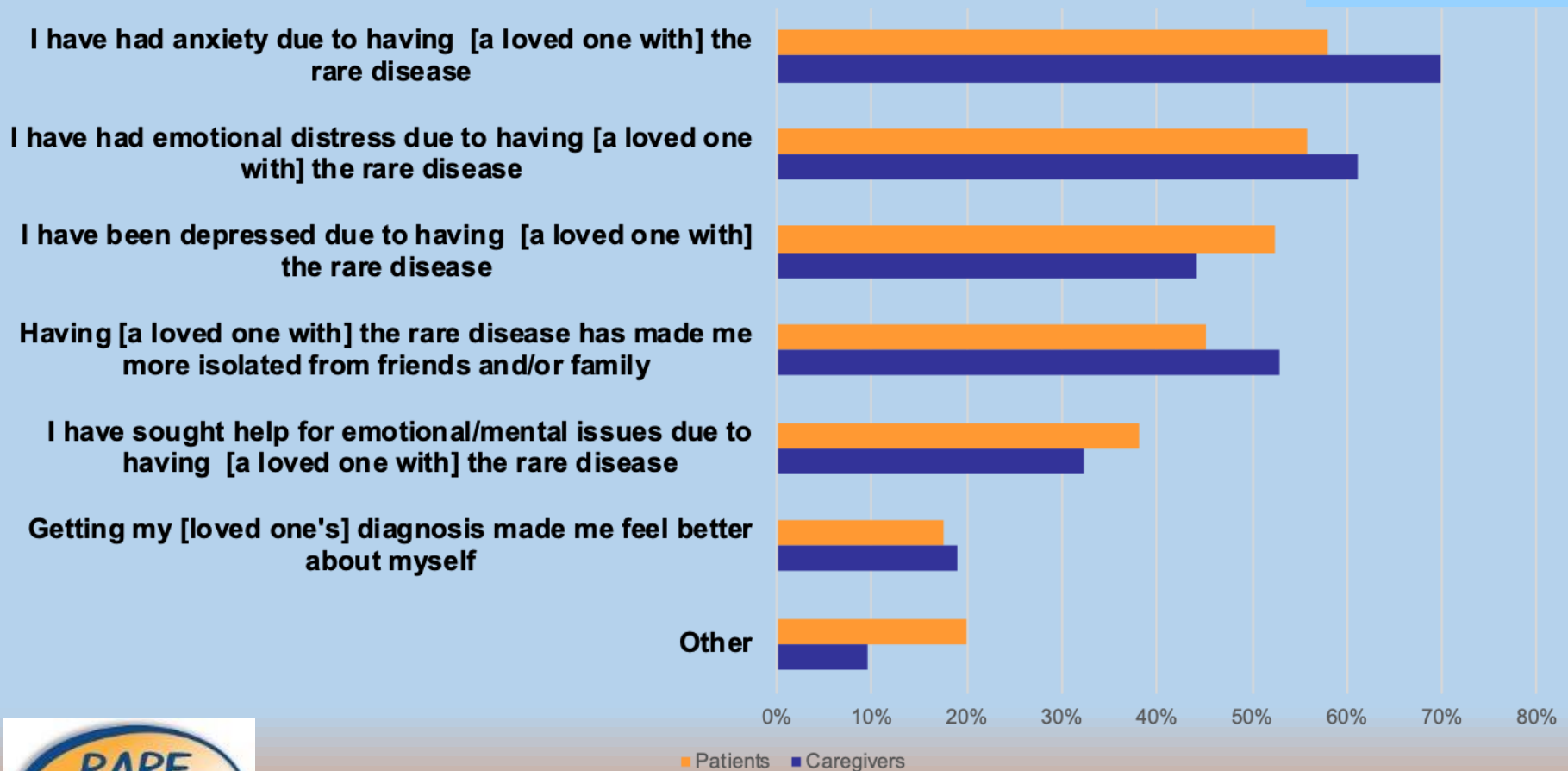
Impact on Family of Rare Disease



A majority of patients and caregivers are affected emotionally by the rare disease.



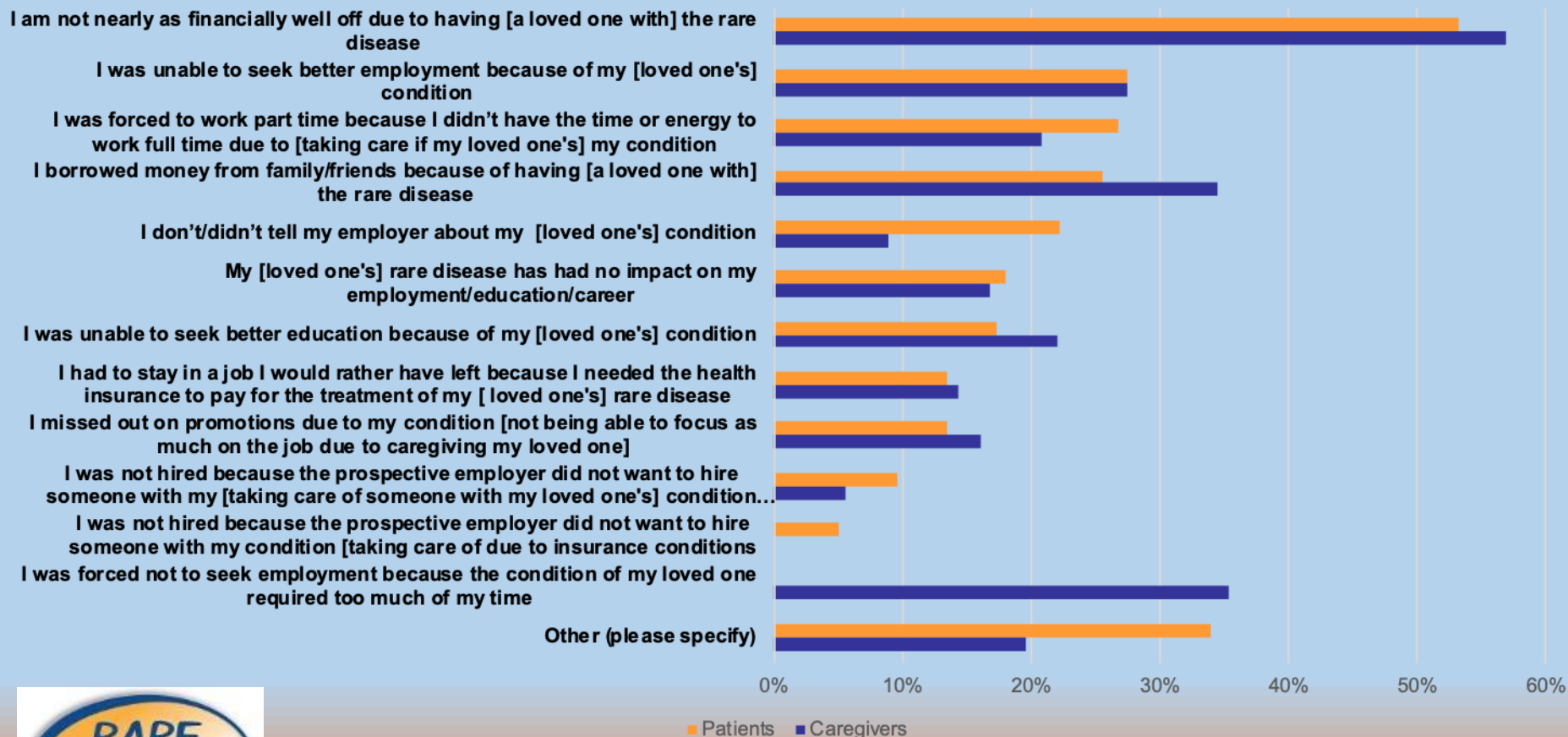
Emotional Impact of Rare Disease



Having the rare disease has a financial impact a majority of families.



Impact on Employment/Education/Financial



Social interaction has been affected by a large majority of patients and caregivers.



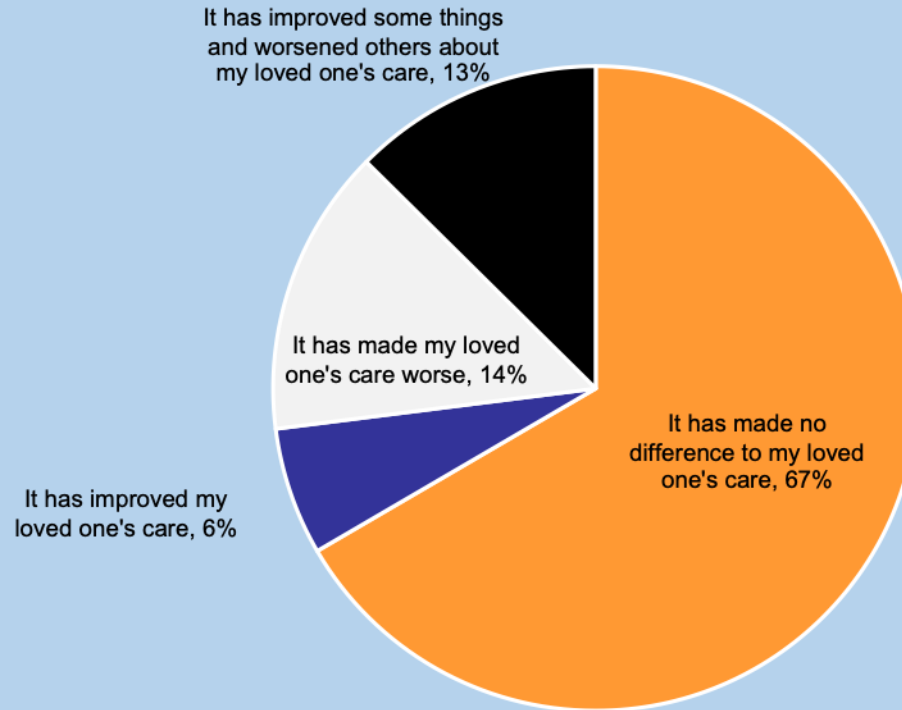
Social Interaction Impact of Rare Disease



Few patients/caregivers indicate that the ACA has improved their care.



Impact of Affordable Care Act



Patient Comments

This disease
steals your
freedom

It has brought me closer to
my daughter who is my
caregiver. I can no longer do
most of the things I enjoyed
doing. But I am doing much
better then a year ago thanks
to my Doctor.

My disease
has changed
our family's
normal.

My family has had to take me to
several doctors to find the diagnosis
and get where we needed to be. They
had missed work with out getting paid.
I was unable to get hired because of
my condition and was unemployed
and did not qualify for disability until
after 12 months of being disabled. We
have had to have fundraiser a to help
raise money to get the medical places I
needed to be in several different
states.

It's been a roller coaster. I live day to
day. I work full time at a hospital
caring for others doing 12 hour
shifts...then on days off I sleep and
miss out on my kids events. It's
tough to explain to my 5 and 6 year
old that mommy's body doesn't work
right and even though I take
medicine all the time it doesn't
always help.

I have spent the past 4 years
searching for a surgeon to
repair issues related to my
rare disease. It has impacted
my entire day to day life. My
kids miss out on a lot because
of my condition.

Can't enjoy any family
gathering, especially
holidays as it hard to
travel and the worry
about what if
something happens
and I'm away from my
regular healthcare
provider



Patient Comments

My entire family has had to take a crash course in understanding everything about the disease. The most difficult part for my parents was coming to terms with there being no cure for the disease.

It has changed the dynamic of my marriage. My husband's role is now my caretaker. My children hear I'm sorry I can't way too often.

Mother of triplets unable to provide the care necessary for my children, I feel like I'm stealing their childhood.

It's hard for others (even family) to even want to know more about the disease, let alone educate themselves. It's hard to educate them unless I feel like I'm pushing it on them, so I just don't

It has taken my life away. At least the life I was used to. This disease impacts my family just as much as it does me. I used to be the mom that did everything and helped with everything. Now I'm lucky if I can at least attend the events.

Life is difficult but we stick together and everything always works out!

It has changed every aspect of our lives. Every single one.



Caregiver Comments

As a parent I have always had to be extremely cognizant of the effect on her siblings. We've had to make sure they don't ever feel neglected and also deal with how they may be handling things emotionally. It's a whole other set of worries in addition to our worry and care for our daughter.

Our lives revolve around son's illness, there is no time or money for anything else.

I've had to pursue a graduate degree for the therapy our loved one needs, because no providers are in our area, nor will any come here..

Despite all the obstacles - we wouldn't trade the rare disease for all the money in the world. We hate it, but our loved one and our family wouldn't be the same without it. We really are closer and stronger because of it.

My husband and I feel that friends and family treat us differently since our daughter's diagnosis. We also feel that family and friends avoid talking about our daughter and her needs

Being told our daughter has CF by my pediatrician 10 days after her birth was the worst day of our lives. There is a general sadness that never goes away. No matter how well other things are going in our lives this is a heavy weight on us. It is hard to accept having something that can't be fixed or cured. Every prayer, birthday wish, etc. is for a cure. I'd give up anything and everything for a cure.

I am now my husband's parent, in effect, as he can't take care of his personal needs, or prepare proper meals, or assist with household chores and lawn care. Everything is my responsibility.



Caregiver Comments

By just looking at my son, one might not "see" anything wrong with him. It's been frustrating with family & friends to where they question if there's anything real "wrong" with him. We find ourselves having to educate them over & over. My son becomes depressed after doctors visits because there's really nothing they can "do" for him. Emotionally the toll is high. It's hard to dream of the future when the future is so unsure. Thanks for asking...

Disease causes social implications more than anything else. More difficult to eat out, go on school field trips that involve meals, not invited to friends' homes as much as other girls in class, not invited to as many parties. This is a big deal for a young girl.

Have to remember to care for ourselves and our older children do we don't burn out or fall apart.

Every single aspect of our day and life is based off of how our son is doing, what season it is and how many machines we need. We spend more time in the home than outside the home. His disease also affects his education. All of the schools we have come into contact with have no idea how to teach or deal with him.

I think it makes it so hard when it is rare because no one understands or is familiar with things so it's a lot of explaining and re explaining what is going on.



Summary

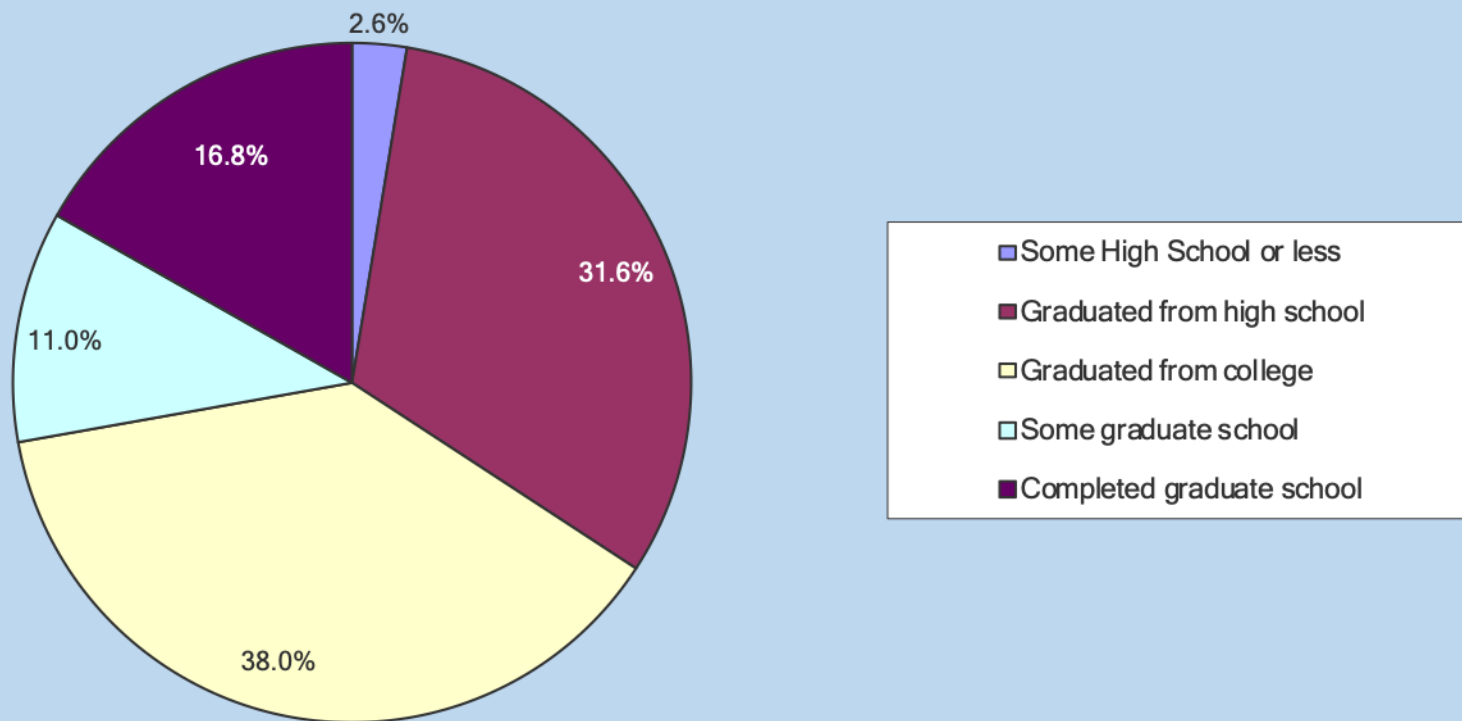
- Rare diseases affect many facets of the lives of patients and caregivers.
- The time required for dealing with their loved one's disease takes away from personal and family activities and interactions.
- The emotional impact of rare disease is particularly high, with relatively more caregivers feeling anxiety, and more patients feeling depressed.
- The financial status of many families has been negatively affected, forcing many patients and caregivers to forego employment and educational opportunities, and to work part-time instead of full time.
- Lack of rare disease knowledge by many healthcare professionals and lack of awareness by others is a source of frustration to many patients and caregivers.
- Most patients and caregivers don't feel the Affordable Care Act has made a difference in their care.



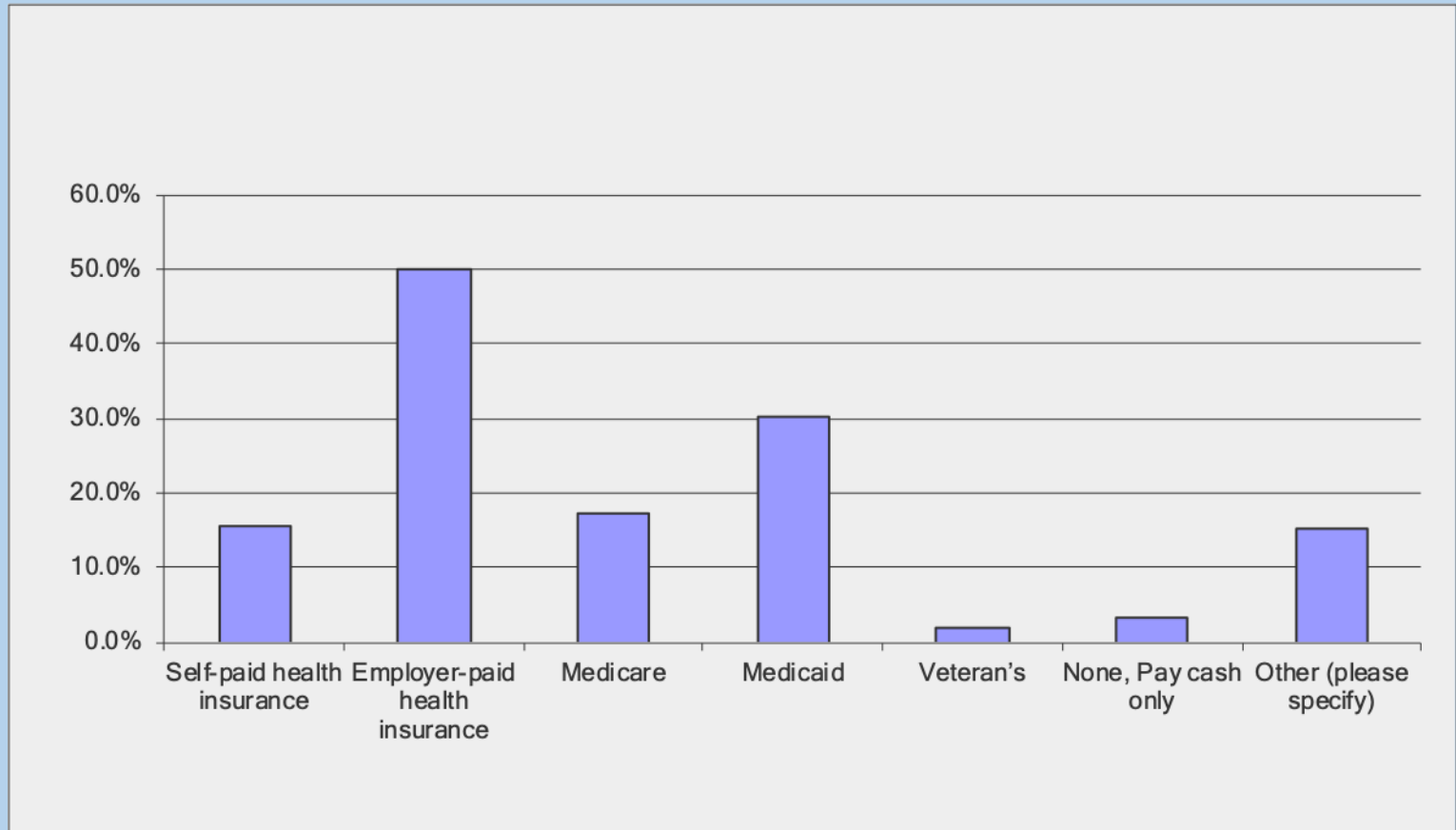
Appendix



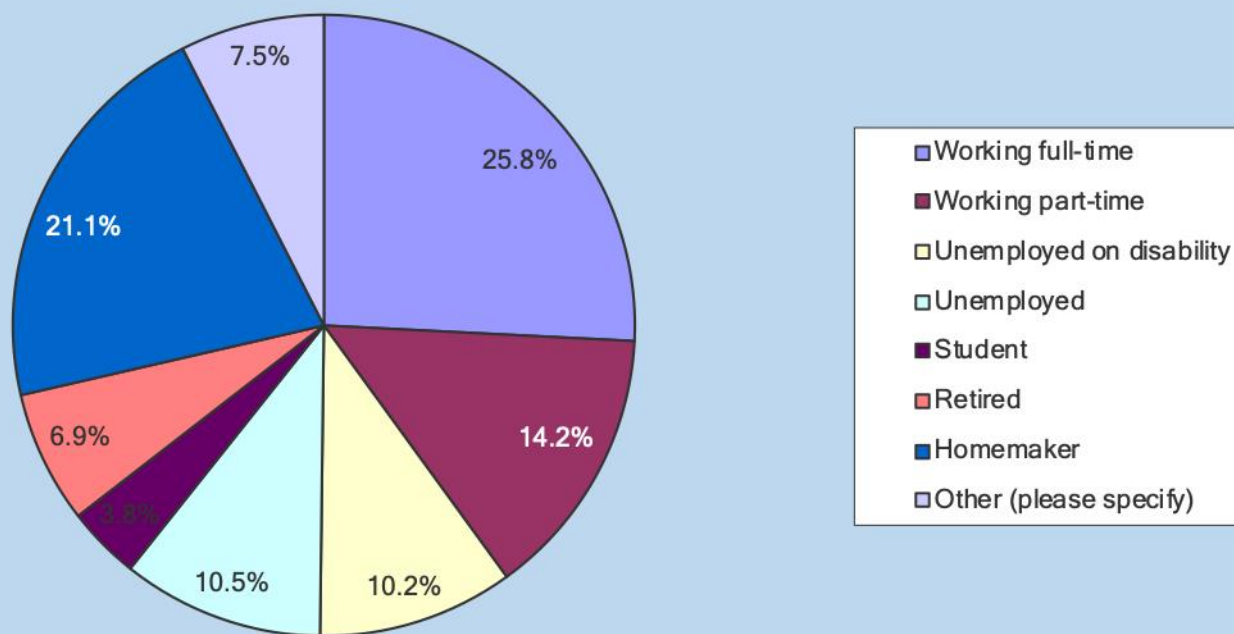
Educational Level



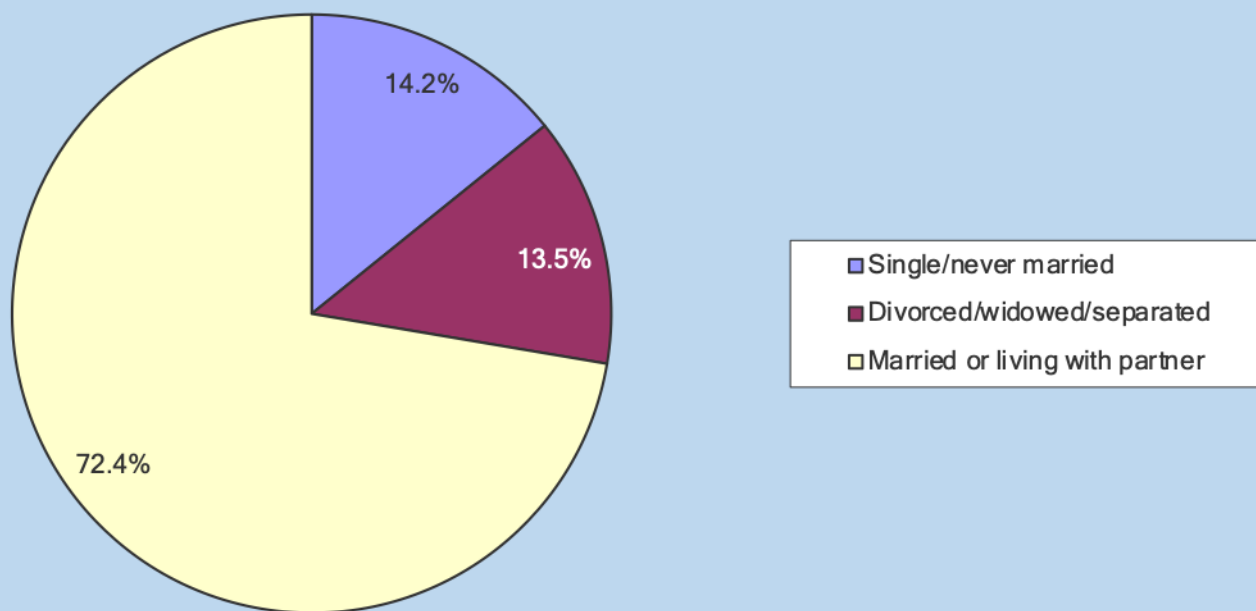
Insurance Coverage



Employment



Marital Status



Household Income

