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DANCING WITH OUR HANDS TIED: AN IMBALANCED FOCUS ON DRUGS FOR ORPHAN DISEASE AND RESEARCH

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**DANCING WITH OUR HANDS TIED: AN
IMBALANCED FOCUS ON DRUGS FOR
ORPHAN DISEASE AND RESEARCH**

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INTRODUCTION

Current efforts to foster research on orphan diseases are focused largely on pharmaceutical treatments. Currently, there are roughly 770 FDA approved pharmaceutical treatments designated for orphan diseases.¹ The number of treatments has greatly improved and it can largely be attributed to government efforts to foster research on orphan diseases. However, more can be done to bridge the gap between the number of available treatments, both pharmaceutical and non-pharmaceutical, and the number of different orphan diseases. These efforts should be supplemented by initiatives to bolster diffusion and commercialization of innovations made by the user-innovator community.

¹ The number of FDA approved treatments for orphan diseases was collected from the U.S. Food & Drug Administration orphan drug designation search. To determine the number of approved treatments, I ran the search for only approved products. The search was run as of March 11, 2019. *Search Orphan Drug Designations and Approvals*, FDA, <https://www.accessdata.fda.gov/scripts/opdlisting/ood/index.cfm> [<https://perma.cc/X64K-2J3E>] (enter “03/11/2019” in “End Date” field and “Only approved products” in “Search results” field) [hereinafter *Designations and Approvals*].

Part I of this note introduces orphan diseases. Part II of this note examines current efforts to foster research on orphan diseases, including the Orphan Drug Act and the U.S. patent system. Through analysis of the incentive provided by these efforts, I conclude that current efforts focus largely on pharmaceutical treatments. Part III of this note introduces user innovation theory and current examples of user innovation by orphan disease patients. In this part, I will show that patient innovation frequently occurs within the orphan disease space. I will also show that, although patients innovate, they do not share or diffuse their innovation enough to maximize the impact of user innovation on orphan diseases. Part VI of this note addresses the successes and challenges of current efforts to foster orphan disease research. Part V of this note makes suggestions for government intervention to incentivize diffusion of patient innovation.

I. ORPHAN DISEASES

An orphan disease, also known as a rare disease, is “any disease or condition which . . . affects less than 200,000 persons in the United States.”² There may be upwards of 7,000 different orphan diseases.³ Some orphan diseases are familiar, like cystic fibrosis and Lou Gehrig’s disease, while others are less familiar.⁴ Most orphan diseases are inherited through gene mutations.⁵ However, environmental factors, such as diet or exposure to chemicals, can also play a role in an orphan disease.⁶ Although each individual orphan disease affects a small amount of the population, in sum, orphan diseases affect millions of people.⁷ It is estimated that twenty-five to thirty million Americans are affected by an orphan disease.⁸ However, not all types of orphan diseases are tracked upon diagnosis.⁹ Thus, the number of cases of orphan diseases in the United States may be lower or higher than the estimation.

The discrepancy between the number of identified orphan diseases and the number of FDA orphan disease designated treatments suggests a

² 21 U.S.C. § 360ee(b)(2) (2017).

³ *FAQs About Rare Diseases*, NIH, <https://rarediseases.info.nih.gov/diseases/pages/31/faqs-about-rare-diseases> [https://perma.cc/WKX3-H5ZQ].

⁴ *Orphan Products: Hope for People with Rare Diseases*, FDA, <https://www.fda.gov/Drugs/ResourcesForYou/Consumers/ucm143563.htm> [https://perma.cc/5RK7-WGAJ] [hereinafter *Orphan Products*].

⁵ *Id.*

⁶ *FAQs About Rare Diseases*, *supra* note 3.

⁷ *Orphan Products*, *supra* note 4.

⁸ *FAQs About Rare Diseases*, *supra* note 3.

⁹ *Id.*

large need for increased research and development of treatment options in this area.¹⁰

II. CURRENT EFFORTS TO FOSTER RESEARCH ON ORPHAN DISEASES

Current efforts to foster research on orphan diseases are largely focused on pharmaceutical treatment. Two formal incentive structures exist to foster research on orphan diseases: the Orphan Drug Act and the patent system. The Orphan Drug Act incentivizes orphan disease drug development by reducing development costs and lengthening exclusivity on the market.¹¹ The U.S. patent system provides a monopoly to inventors to recoup costs on research and development.

A. ORPHAN DRUG ACT

In 1983, President Ronald Reagan signed the Orphan Drug Act into law.¹² Congress made several findings for the Orphan Drug Act:

- (1) there are many diseases and conditions . . . which affect such small numbers of individuals . . . that the diseases and conditions are considered rare in the United States;
- (2) adequate drugs for many of such diseases and conditions have not been developed;
- (3) drugs for these diseases and conditions are commonly referred to as “orphan drugs”;
- (4) because so few individuals are affected by any one rare disease or condition, a pharmaceutical company which develops an orphan drug may reasonably expect the drug to generate relatively small sales in comparison to the cost of developing the drug and consequently to incur a financial loss;
- (5) there is reason to believe that some promising orphan drugs will not be developed unless changes are made in the applicable Federal laws to reduce the costs of developing such drugs and to provide financial incentives to develop such drugs; and
- (6) it is in the public interest to provide such changes and incentives for the development of orphan drugs.¹³

¹⁰ There are only 668 FDA designated and approved orphan drug treatments. *Compare Designations and Approvals*, *supra* note 1, with *FAQs About Rare Diseases*, *supra* note 3.

¹¹ See Orphan Drug Act, Pub. L. No. 97-414, 96 Stat. 2049 (1983).

¹² Sinead M. Murphy et al., *Unintended Effects of Orphan Product Designation for Rare Neurological Diseases*, 72 ANN. NEUROL. 481, 481 (2012).

¹³ Orphan Drug Act, *supra* note 11 at § 1 (emphasis added).

The intended purpose of the Orphan Drug Act, therefore, was to incentivize orphan drug development by reducing costs and providing incentives.¹⁴ To accomplish this goal, the Orphan Drug Act provides for market exclusivity, tax credits, access to research grants, and fee waivers.¹⁵

To gain access to the incentives provided by the Orphan Drug Act, a drug must be designated as a drug for a rare disease or condition.¹⁶ To be designated as a drug for a rare disease or condition, a drug sponsor must submit an application detailing its application to a rare disease or condition.¹⁷

The Orphan Drug Act grants seven years of market exclusivity to an approved application for a drug designated for a rare disease or condition.¹⁸ On average, this period of seven-year exclusivity is two years longer than the typical period of exclusivity for a drug.¹⁹ The period of exclusivity is subject to two exceptions.²⁰ If the holder of the exclusive approval “cannot ensure the availability of sufficient quantities of the drug to meet the needs of persons with the disease or condition for which the drug was designated,” another application for a drug that is the same may be granted.²¹ Additionally, another application for a drug that is the same may be granted if the holder of the exclusive approval consents to the approval.²²

The Orphan Drug Act provides a twenty-five percent tax credit on research and development costs for orphan disease drugs.²³ The tax credit

¹⁴ *Id.*

¹⁵ See 21 U.S.C. § 360 (2017); 26 U.S.C. § 45C (2017).

¹⁶ See 21 U.S.C. § 360bb (2017).

¹⁷ *Id.* For example, the drug Kymriah was designated in 2015 as an orphan drug for the treatment of Acute Lymphoblastic Leukemia, a rare disease, following Novartis’s application for designation. *Designations and Approvals, supra* note 1.

¹⁸ 21 U.S.C. § 360cc (2017). The term of a patent is twenty years from the date of filing the patent application. *Patents and Exclusivity*, FDA, (May 19, 2015) <https://www.fda.gov/downloads/drugs/developmentapprovalprocess/smallbusinessassistance/ucm447307.pdf> [<https://perma.cc/D9x4-M6S9>]. Exclusivity, on the other hand, is granted by the FDA upon approval of the drug. *Id.* Exclusivity can run concurrent with the patent term and may, in some cases, extend past the patent period. *Id.* For example, if a patent is filed upon drug discovery, only part of the twenty-year period will remain after research and development. Exclusivity can lengthen this period if less than seven years remain on the patent term. *Id.*

¹⁹ Ed Rensi, *The Orphan Drug Act Has Been a Huge Success*, WALL ST. J. (Jun. 23, 2008, 12:01 AM), <https://www.wsj.com/articles/SB121417838559995535> [<https://perma.cc/6C6V-2D43>].

²⁰ 21 U.S.C. § 360cc.

²¹ *Id.*

²² *Id.*

²³ 26 U.S.C. § 45C (2017).

only applies to costs incurred before the drug approval.²⁴ In addition, the costs incurred are only taken into account “only to the extent such testing is related to the use of a drug for the rare disease or condition for which it was designated.”²⁵

The Orphan Drug Act also grants the Secretary of Health and Human Services the power to make grants and enter into contracts “to assist in defraying the costs of developing drugs for rare diseases or conditions, including qualified testing expenses.”²⁶ Qualified testing includes “observational studies . . . conducted to assist in the understanding of the natural history of a rare disease or condition.”²⁷ Qualified testing also includes observational studies to “develop or validate a drug development tool” and to “understand the full spectrum of the disease manifestations.”²⁸ For each year between 2013 and 2017, \$30 million was appropriated for grants.²⁹ The statute has been reinstated for the same amount of funding for each year between 2018 and 2022.³⁰

Finally, the Orphan Drug Act provides for priority review for treatments directed to rare pediatric diseases.³¹ Since the passage of the Orphan Drug Act, the FDA has approved more than 600 orphan drug designations.³² Compared to only ten FDA-approved treatments for orphan diseases prior to the passage of the Orphan Drug Act, the Act can be marked as a success.³³

The Orphan Drug Act creates new incentives for the FDA drug approval process. Pharmaceutical companies seeking to sell a new prescription drug on the market must go through FDA approval.³⁴ The FDA approval process requires intensive testing. First, laboratory and animal tests must be completed.³⁵ Next, the drug must be tested on humans

²⁴ *Id.*

²⁵ *Id.*

²⁶ 21 U.S.C. § 360ee.

²⁷ *Id.*

²⁸ *Id.*

²⁹ *Id.*

³⁰ FDA Reauthorization Act of 2017, Pub. L. No. 115-52, § 603, 131 Stat. 1005, 1048 (2017).

³¹ 21 U.S.C. § 360ff (Supp. V 2012).

³² *FDA's Orphan Drug Modernization Plan*, FDA, 2 (June 29, 2017), <https://www.fda.gov/downloads/ForIndustry/DevelopingProductsforRareDiseasesConditions/HowtoapplyforOrphanProductDesignation/UCM565068.pdf> [<https://perma.cc/B69Q-2MSL>] [hereinafter *Modernization Plan*].

³³ *Id.*

³⁴ *Development & Approval Process (Drugs)*, FDA (June 13, 2018), <https://www.fda.gov/Drugs/DevelopmentApprovalProcess/default.htm> [<https://perma.cc/HM7H-4EYS>] [hereinafter *FDA Approval Process*].

³⁵ *Id.*

“determine whether the drug is safe when used to treat a disease.”³⁶ Subsequently, the company must send an application to the FDA with the test results, manufacturing information, and a proposed label.³⁷ This process is long and expensive. New drug approval in the United States requires, on average, twelve years and one billion dollars.³⁸

While the FDA approval process is fruitful for drugs with a high probability of commercial success and market reach, sales of approved drugs and medical devices aimed at small markets may not produce enough revenue to justify the time and monetary investment required for FDA approval. In the case of orphan diseases, the failure of the commercial market is largely attributable to small market size.³⁹ Because each disease affects such a small population, there is no way to expand the market for each disease.⁴⁰ Small market size “makes it commercially unattractive for pharmaceutical firms and other medical suppliers to invest in developing new products specifically for rare diseases.”⁴¹ Thus, pharmaceutical firms and medical suppliers generally do not invest in orphan diseases, leaving them underserved by the traditional model of FDA approval and commercialization. The Orphan Drug Act remedies some of these problems with monetary incentives and market exclusivity.

Although the Orphan Drug Act positively affects the amount of drugs targeted towards orphan disease treatment, the wide array of orphan diseases necessarily means that not all orphan disease research will receive adequate funding. Priority review for pediatric-focused treatments, while incentivizing for some, excludes orphan diseases that do not primarily affect individuals under the age of eighteen.⁴² What is left is the seven-year exclusivity provision and the tax credit that will apply to all approved drugs targeted at an orphan disease.

³⁶ *Id.*

³⁷ *Id.*; Gail A. Van Norman, *Drugs, Devices, and the FDA: Part 1*, 1 JACC: BASIC TO TRANSLATIONAL SCI. 170, 178 (2016) [<https://perma.cc/7DKW-H4U4>].

³⁸ Van Norman, *supra* 37 at 178.

³⁹ Daron Acemoglu & Joshua Linn, *Market Size in Innovation: Theory and Evidence from the Pharmaceutical Industry*, 119 Q. J. ECON. 1049, 1049–90 (2004) [<https://perma.cc/7LE6-AZSH>].

⁴⁰ *Id.*

⁴¹ Pedro Oliveira et al., *Innovation by Patients with Rare Diseases and Chronic Needs*, 10 ORPHANET J. RARE DISEASES 1, 4 (2015).

⁴² 21 U.S.C. § 360ff (Supp. V 2012).

B. PATENT SYSTEM

The United States Patent and Trademark Office (USPTO) issues property rights, in the form of a patent, to inventors.⁴³ A patent grants the inventor “the right to exclude others from making, using, offering for sale, or selling the invention.”⁴⁴ The patent owner can exercise these rights for twenty years following the date of the patent application.⁴⁵ Infringement occurs when another “without authority makes, uses, offers to sell, or sells any patented invention.”⁴⁶ If a patent is infringed, the owner is entitled to relief.⁴⁷ This relief can come in the form of an injunction or damages.⁴⁸ Patent rights are available to any inventor who submits an application to the USPTO and whose invention meets the requirements for patent-eligibility.⁴⁹ New and novel pharmaceutical treatments for orphan diseases likely meet the requirements for patent-eligibility.⁵⁰ Thus, the U.S. patent system may also be skewed toward incentivizing pharmaceutical treatments.

III. USER INNOVATION BY PATIENTS

User innovation theory centers around the theory that users can and will develop the technology they need.⁵¹ There are many examples of user innovation by patients with orphan diseases, including an ostomy patient who developed technology that warns “patients when their ostomy bags are full so they can empty them without risking overflow.”⁵² In this example, we can see that the innovation was driven by what the end-user, in this case the patient, needed based on his own experience with the disease.⁵³

⁴³ *General information concerning patents*, USPTO (Oct. 2015), <https://www.uspto.gov/patents-getting-started/general-information-concerning-patents> [<https://perma.cc/ED6Z-7YT6>] [hereinafter *General Patent Information*].

⁴⁴ 35 U.S.C. § 154 (2017).

⁴⁵ *Id.*

⁴⁶ *Id.* § 271.

⁴⁷ *General Patent Information*, *supra* note 43.

⁴⁸ *Id.*

⁴⁹ *Id.*

⁵⁰ Angélique McCall & Gene Quinn, *The FDA Process, Patents and Market Exclusivity*, IPWATCHDOG (Mar. 12, 2017), <http://www.ipwatchdog.com/2017/03/12/fda-process-patents-market-exclusivity/id=79305/> [<https://perma.cc/A6CS-DWA9>].

⁵¹ ERIC VON HIPPEL, *DEMOCRATIZING INNOVATION* 1 (2005).

⁵² Helena Canhão et al., *Revolutionising Healthcare by Empowering Patients to Innovate*, 1 EMJ INNOV. 31, 32 (2017).

⁵³ *Id.*

C. INTRODUCTION TO USER INNOVATION THEORY

The main justification for patent law is utilitarian in nature.⁵⁴ The “purpose of the patent system is to induce the creation and commercialization of technology that otherwise would be easily appropriated.”⁵⁵ Thus, “only those inventions that would otherwise not materialize, or would be discovered only after a longer passage of time, should receive the benefits of patent protection.”⁵⁶

Technological innovations are public goods.⁵⁷ Public goods can be replicated and they are nonrivalrous, meaning that “enjoyment of them by one person does not prevent enjoyment of them by other persons.”⁵⁸ Because they are nonrivalrous, “other people will be able to take advantage of [the innovations] for free.”⁵⁹ These nonpaying consumers are often called “free riders.”⁶⁰ Without intervention, innovators will not disclose their innovations because “innovators will be unable to recoup the costs of their innovations.”⁶¹ The fear is that free-riding behavior, without intervention, would lead to “significant underinvestment in . . . intellectual resources because of the risk that competitors would appropriate their value.”⁶²

One solution to the free-rider problem is to confer intellectual property rights upon innovators.⁶³ As discussed above, the USPTO grants an inventor “the right to exclude others from making, using, offering for sale, or selling the invention.”⁶⁴ These property rights “lower the costs of exclusion, enable transactions, and mitigate the risks to investment posed by free riders.”⁶⁵ Although intellectual property rights may resolve the free-rider problem, there are other factors that may mitigate the need for intellectual property rights to stimulate innovation.

⁵⁴ Alan Devlon & Neel Sukhatme, *Self-Realizing Inventions and the Utilitarian Foundation of Patent Law*, 51 WM. & MARY L. REV. 897, 901 (2009).

⁵⁵ *Id.*

⁵⁶ *Id.*

⁵⁷ William Fisher, *Intellectual Property and Innovation: Theoretical, Empirical, and Historical Perspectives*, 37 BELEIDSSTUDIES TECHNOLOGIE ECONOMIE 47, 47 (2001), <https://cyber.harvard.edu/people/tfisher/Innovation.pdf> [<https://perma.cc/GU7F-6RGD>].

⁵⁸ *Id.* at 47.

⁵⁹ *Id.*

⁶⁰ BRETT M. FRISCHMANN ET AL., *Governing Knowledge Commons*, in GOVERNING KNOWLEDGE COMMONS 1, 6 (Brett M. Frischmann et al. eds., 2014).

⁶¹ Fisher, *supra* note 57, at 47.

⁶² FRISCHMANN ET AL., *supra* note 60, at 6 (internal citation omitted).

⁶³ Fisher, *supra* note 57, at 49.

⁶⁴ 35 U.S.C. § 154 (2017).

⁶⁵ FRISCHMANN ET AL., *supra* note 60, at 7.

Even in the presence of free riders, intrinsic user motivations for innovation may be sufficient to stimulate innovation without conferring property rights to innovators.⁶⁶ Under the user innovation theory, “user innovation is innovation motivated by an intention to use . . . an innovative technology.”⁶⁷ User innovators are still in part motivated by utilitarian values, as they “expect to be rewarded by the use value of what they are creating . . . and . . . intrinsically by such things as the fun and learning experience derived from creating it.”⁶⁸ Ideas for inventions come from users who develop improvements to serve their own needs.⁶⁹ User innovation thrives when users have “unique local information about their needs and the technical capacity to make inventions that fulfill those needs.”⁷⁰

User innovation theory depends on users of existing technology. These users are motivated by developing technology for their personal use.⁷¹ User innovators “are unique in that they alone benefit directly from innovations.”⁷² Because user innovations benefit the user, “user innovations will . . . tend to be those that leverage the user’s information advantages . . . rather than those that leverage manufacturers’ information advantages.”⁷³

User innovators will invest in developing innovations because they “can develop exactly what they want, rather than relying on manufacturers to act as their own (often very imperfect) agents.”⁷⁴ Additionally, user innovators “derive benefits from developing and using their inventions, which motivate[s] them to invest the effort necessary for invention.”⁷⁵

Even though user innovators must invest personal funds and time in developing an innovation, many user innovators “‘freely reveal’ their innovations to others because of private benefits they are able to obtain as a result.”⁷⁶ When a user freely reveals their invention, “all intellectual property rights to that information are voluntarily given up by the

⁶⁶ *Id.* at 10.

⁶⁷ Katherine J. Strandburg, *Users as Innovators: Implications for Patent Doctrine*, 79 U. COLO. L. REV. 467, 478 (2008).

⁶⁸ Ruth Maria Stock et al., *Impacts of Hedonic and Utilitarian User Motives on the Innovativeness of User-Developed Solutions*, 32 J. PROD. INNOVATION MGMT. 389, 389 (May 2015).

⁶⁹ VON HIPPEL, *supra* note 51, at 1.

⁷⁰ Katherine J. Strandburg, *Accommodating User Innovation in the International Intellectual Property Regime: A Global Administrative Law Approach*, 2009 ACTA JURIDICA 283, 293–94 (2009).

⁷¹ Strandburg, *supra* note 67, at 469.

⁷² VON HIPPEL, *supra* note 51, at 3.

⁷³ *Id.* at 45–61.

⁷⁴ *Id.* at 1.

⁷⁵ Strandburg, *supra* note 67, at 469.

⁷⁶ *Id.* at 469–70.

innovator, and all interested parties are given access to it.”⁷⁷ In other words, the innovation becomes a public good.⁷⁸ A user innovator is free to reveal their innovation because they “receive[] sufficient ‘return on investment’ from developing and using the invention to compensate for expenditures on developing it.”⁷⁹ The user innovator’s return on investment comes in many forms. For example, monetary compensation for use, reputational enhancement, or simple enjoyment of using the invention or of the inventive process.⁸⁰

Some technologies are more suited for user innovation than others.⁸¹ The suitability of a technology with user innovation depends on several factors: “the heterogeneity of uses, the presence of lead users, the technical difficulty of invention in a particular field, and the costs of development.”⁸² In a technology with high heterogeneity of uses, “many individual users or user firms want something different in a product type.”⁸³ A technology with low heterogeneity of uses would already be satisfied through traditional commercial models because “[m]ass manufacturers tend to follow a strategy of developing products that are designed to meet the needs of a large market segment well enough to induce purchase from and capture significant profits from a large number of customers.”⁸⁴ High heterogeneity of uses also leads to high willingness to pay to get what you want, which lends well to user innovation theory.⁸⁵

Lead users are users who are “ahead of the majority of users in their populations with respect to an important market trend.”⁸⁶ Lead users “expect to gain relatively high benefits from a solution to the needs they have encountered there.”⁸⁷ Because lead users are ahead of the market, they are more likely to make improvements to technology before the market. This also means that their needs are likely not represented on the market. Finally, because they expect to gain high benefits, they are more likely to incur the costs of developing the innovation on their own.

For user innovator communities where social norms are informally enforced, rational choice theory explains how collective action problems

⁷⁷ VON HIPPEL, *supra* note 51, at 9.

⁷⁸ *Id.*

⁷⁹ Strandburg, *supra* note 67, at 478.

⁸⁰ *Id.*

⁸¹ Strandburg, *supra* note 70, at 294.

⁸² *Id.*

⁸³ VON HIPPEL, *supra* note 51, at 33.

⁸⁴ *Id.* at 5.

⁸⁵ *Id.* at 5–6.

⁸⁶ *Id.* at 4.

⁸⁷ *Id.*

can be solved.⁸⁸ The stability and viability of sharing within these communities depends on:

- (1) the fact that the norm is beneficial for community members in light of their preferences;
- (2) the community's ability to detect defections from the norm; and
- (3) the community's ability to impose penalties that are sufficient to deter defection yet not so costly to impose that they overwhelm the coordination benefit.⁸⁹

D. USER INNOVATION IN ORPHAN DISEASES

Patients who engage in user innovation are motivated by the prospect of curing or improving their own disease. With this in mind, we can see how each patient innovation caters to their own individual motivations. Several studies have shown that orphan disease patients engage in innovation to create solutions to their own medical needs.⁹⁰ For example, in one study, thirty-six percent of a sample of rare disease patients and their non-professional caregivers developed innovations to improve the management of their disease.⁹¹ In a more narrowly directed study, researchers found that fifty percent of the solutions for patients with cystic fibrosis were developed by patients.⁹²

These user innovations range from simple tools for everyday use to highly sophisticated solutions.⁹³ For example, a daughter of a patient with dementia swapped colorful plates for white plates to minimize distraction while her father was eating.⁹⁴ In another example, a mother of a patient with Angelman syndrome filled a room with floating balloons to encourage her child to jump and reach for the balloons, greatly increasing his physical abilities.⁹⁵ Both examples are arguably simple tools for everyday use. Patients have also developed more sophisticated innovations. For example, a patient with Myasthenia gravis developed "a metal two-hook button aid that helps her button pants without the assistance of others."⁹⁶ In another example, a Marfan syndrome patient developed a textile mesh support for

⁸⁸ Katherine J. Strandburg, *User Innovator Community Norms: At the Boundary between Academic and Industry Research*, 77 *FORDHAM L. REV.* 2237, 2242 (2009).

⁸⁹ *Id.*

⁹⁰ See Canhão, *supra* note 52, at 32; Oliveira *supra* note 41, at 6.

⁹¹ Oliveira, *supra* note 41, at 4.

⁹² Canhão, *supra* note 52, at 31.

⁹³ Oliveira, *supra* note 41, at 2.

⁹⁴ Canhão, *supra* note 52, at 32.

⁹⁵ Oliveira, *supra* note 41, at 4.

⁹⁶ *Id.* at 5.

dilated heart aorta.⁹⁷ From these selected examples, we can see that user innovation occurs within the context of patients with orphan diseases.

E. LACK OF DIFFUSION BY PATIENT INNOVATORS

Although patients with orphan diseases engage in successful user innovation, very few of these solutions are shared.⁹⁸ In a study conducted by Pedro Oliveira, only thirty percent of the sample of orphan disease innovators reported sharing their solution in some way.⁹⁹ Among those innovators who shared their solution, the vast majority shared it with other patients, but few showed it to medical professions, shared the information on a website or blog, shared it through media, showed it to commercial entities, spent time or money to help others use the solution, or made a manual that helps using the solution.¹⁰⁰

There are several reasons why patients may not share their innovations. For example, some may not have financial incentives or opportunities to enter the process of approval or commercialization.¹⁰¹ However, financial incentives may not explain the entirety of the issue. Oliveira found that the “strongest predictor of information sharing was the observed difference in the respondents’ overall quality of life before and after using a solution.”¹⁰² If the solution does not improve quality of life, a patient may not feel that it is beneficial to share—if it didn’t help them, why would it help anyone else?

IV. CURRENT EFFORTS’ SUCCESS AND CHALLENGES

F. ORPHAN DRUG ACT

The Orphan Drug Act is viewed as a huge success by many. “[I]n the decade before the law was passed only 10 new drugs for rare diseases were developed.”¹⁰³ Now, there are roughly 770 FDA designated and approved orphan drug treatments.¹⁰⁴

As previously discussed, the FDA provides orphan drug designation for drugs targeted towards orphan diseases.¹⁰⁵ Just from 2000 to 2009, over

⁹⁷ *Id.* at 2.

⁹⁸ Canhão, *supra* note 52, at 32.

⁹⁹ Oliveira, *supra* note 41, at 5.

¹⁰⁰ *Id.* at 7.

¹⁰¹ Canhão, *supra* note 52, at 33.

¹⁰² Oliveira, *supra* note 41, at 6.

¹⁰³ Rensi, *supra* note 19.

¹⁰⁴ *Designations and Approvals*, *supra* note 1.

¹⁰⁵ See 21 U.S.C. § 360bb (2017).

one thousand orphan drugs were designated as orphan drugs.¹⁰⁶ With a rise in the number of designation requests, a backlog ensued in the FDA designation system.¹⁰⁷ The FDA's Orphan Drug Modernization Plan, unveiled in June 2017, seeks "to completely eliminate the agency's existing orphan designation backlog and ensure continued timely response to all new requests for designation with firm deadlines."¹⁰⁸ In the first ninety days, around September 2017, the FDA planned to "complete reviews of all orphan drug designations . . . older than 120 days."¹⁰⁹ The FDA is also committed to responding to any subsequent designation requests within ninety days of their receipt.¹¹⁰

Looking at the increase in orphan disease treatments, the Orphan Drug Act has largely been a success. Though the continued success of the Orphan Drug Act will depend on the elimination of the backlog. It is also important to note that the Orphan Drug Act may not do enough even to incentivize drug development, especially in cases where the number afflicted by a certain orphan disease is remarkably low. These diseases may have too small of a market to recoup the costs no matter how long the period of exclusivity is.

The Orphan Drug Act has functioned without serious threats for over thirty years and funding has been appropriated through 2022.¹¹¹ However, like any government policy, it is subject to change or removal. In 2017, in the wake of a new tax plan, Congress weighed repeal of the tax credit for orphan disease drugs.¹¹² Repeal of the orphan drug tax credit would save the government approximately \$54 billion over the next ten years.¹¹³ Although some argued the cut would jeopardize orphan drug development, some argued that "major drugmakers have exploited [the program] by obtaining the orphan designation for billion-dollar blockbuster drugs . . . that were already on the market."¹¹⁴ A new tax plan was enacted, and, for tax years beginning after 2017, the credit rate was cut in half; instead of the

¹⁰⁶ Chandana Thorat et al., *What the Orphan Drug Act Has Done Lately for Children With Rare Diseases: A 10-Year Analysis*, 129 PEDIATRICS 516, 516 (2012).

¹⁰⁷ See *Designating an Orphan Product: Drugs and Biological Products*, FDA, <https://www.fda.gov/ForIndustry/DevelopingProductsforRareDiseasesConditions/HowtoapplyforOrphanProductDesignation/default.htm> [<https://perma.cc/AY2R-6YE8>] [hereinafter *Designating an Orphan Product*].

¹⁰⁸ *Id.*

¹⁰⁹ *Modernization Plan*, *supra* note 32, at 3.

¹¹⁰ *Id.*

¹¹¹ 21 U.S.C. § 360ee (2017).

¹¹² Katie Thomas & Sheila Kaplan, *Congress Weighs Repeal of Tax Credit for Rare Disease Drugs*, N.Y. TIMES (November 8, 2017), <https://www.nytimes.com/2017/11/08/health/republican-tax-plan-orphan-drugs-rare-diseases.html> [<https://perma.cc/B6C9-X8JE>].

¹¹³ *Id.*

¹¹⁴ *Id.*

previous 50% credit for orphan drug testing expenses, filers now receive a 25% maximum credit.¹¹⁵

G. PATENTABILITY CHALLENGES

To be patentable, an invention must meet several requirements. If an invention does not meet each requirement, the patent application will be denied. Not all, but many user innovations will qualify for patent protection.

1. § 103 – THE NOVELTY REQUIREMENT

Patent claims must be non-obvious.¹¹⁶ The relevant statutory provision states: “A patent for a claimed invention may not be obtained . . . if the differences between the claimed invention and the prior art are such that the claimed invention as a whole would have been obvious before the effective filing date of the claimed invention.”¹¹⁷

This statutory provision requires that an innovation be novel to be patentable. Many user innovations within the orphan disease space are not novel.¹¹⁸ Instead, the majority of these innovations are redevelopments—“known to medicine, although not to the patient or caregivers who redeveloped them.”¹¹⁹ This indicates that many user innovations by patients with rare diseases would not be patentable under 35 U.S.C. § 103. In fact, “solution novelty does not necessarily ensure utility for the user.”¹²⁰ If a positive change in disease management or lifestyle motivates a patient to innovate, there may be little incentive to aim for novelty if it does not necessarily lead to utility. If novelty is not a goal of all user innovators within the orphan disease space, something beyond the patent system is required to incentivize innovation and sharing.

2. § 112 – THE ENABLEMENT REQUIREMENT

Patent applications must contain a written description of the invention.¹²¹ The relevant statutory provision states:

¹¹⁵ *Id.*; Zachary Brennan, *Senate, House Agree to Cut Orphan Drug Research Credit in Half in Tax Bill*, REGULATORY AFFAIRS PROFESSIONALS SOCIETY, <https://www.raps.org/regulatory-focus%E2%84%A2/news-articles/2017/12/senate,-house-agree-to-cut-orphan-drug-research-credit-in-half-in-tax-bill> [https://perma.cc/2CZW-ART5].

¹¹⁶ 35 U.S.C. § 103 (2017).

¹¹⁷ *Id.*

¹¹⁸ In a study of 182 rare disease patients who developed an invention, 78% of the solutions were not novel. Oliveira, *supra* note 41, at 4.

¹¹⁹ *Id.* at 6.

¹²⁰ Stock, *supra* note 68, at 393.

¹²¹ 35 U.S.C. § 112.

The specification shall contain a written description of the invention, and of the manner and process of making and using it, in such full, clear, concise, and exact terms as to enable any person skilled in the art to which it pertains, or with which it is most nearly connected, to make and use the same, and shall set forth the best mode contemplated by the inventor. . . .¹²²

A patent claim will be rendered “invalid if it is not supported by an enabling disclosure.”¹²³ The purpose of this requirement is “to ensure that the invention is communicated to the interested public in a meaningful way.”¹²⁴ So long as any person skilled in the art can make and use the invention without undue experimentation, the claimed invention will be enabled.¹²⁵ Patient innovations will meet this requirement as long as the application is sufficiently detailed, regardless of the technology involved.

3. § 101 – THE UTILITY REQUIREMENT

Article I, Section 8 of the United States Constitution granted Congress the power “[t]o promote the progress of Science and the *useful* Arts.”¹²⁶ In turn, under 35 U.S.C. § 101, “an applicant must claim an invention that is statutory subject matter and must show that the claimed invention is ‘useful’ for some purpose”¹²⁷ If an applicant fails to identify specific and substantial utility or “fails to disclose enough information about the invention to make its usefulness immediately apparent to those familiar with the technological field of the invention,” a patent will not meet the useful invention requirement.¹²⁸ To show specific and substantial utility, an applicant must show “that the claimed invention is useful for any particular practical purpose.”¹²⁹ There need only be “one credible assertion of specific and substantial utility for each claimed invention to satisfy the utility requirement.”¹³⁰

Here, the question is whether an improvement in comfort or quality of life is enough to satisfy specific and substantial utility. Not all patient innovations will impact the course of their disease, as “patients may greatly value innovations that may have no impact on the course of their disease,

¹²² *Id.*

¹²³ Manual of Patent Examining Procedure, 9th ed., rev. 8.2017 (Jan. 2018) (hereinafter “MPEP”), § 2164, <https://www.uspto.gov/web/offices/pac/mpep/s2164.html> [<https://perma.cc/4752-2CG7>].

¹²⁴ *Id.*

¹²⁵ *In re Wands*, 858 F.2d 731, 735 (Fed. Cir. 1988).

¹²⁶ U.S. CONST. art. I, § 8, cl. 8 (emphasis added).

¹²⁷ MPEP § 2107, <https://www.uspto.gov/web/offices/pac/mpep/s2107.html> [<https://perma.cc/96YN-DFQM>] [hereinafter Utility Requirement].

¹²⁸ *Id.*

¹²⁹ *Id.*

¹³⁰ *Id.*

but that improve their comfort or other aspects of their quality of life while living with their disease.”¹³¹

First, let’s examine the specific utility of patient innovations. In *In re Fisher*, the Federal Circuit stated that “specific utility” means that the subject matter claimed can “provide a well-defined and particular benefit to the public.”¹³² For example, a “general statement of diagnostic utility, such as diagnosing an unspecified disease, would ordinarily be insufficient absent a disclosure of what condition can be diagnosed.”¹³³ Under this logic, as long as the patient innovator discloses the specific orphan disease applicable to the innovation, the specific utility requirement will be met.

Next, let’s examine the substantial utility of patient innovations. To satisfy the “substantial utility” requirement, “an asserted use must show that the claimed invention has a significant and presently available benefit to the public.”¹³⁴ If the utility “require[s] or constitute[s] carrying out further research to identify or reasonably confirm a ‘real world’ context of use,” it is not a substantial utility.¹³⁵ While a method of treating an unspecified disease or condition does not define “substantial utility” because it would require “carrying out further research to identify or reasonably confirm a ‘real world’ context of use,” a method of treating a specified disease or condition does define “substantial utility.”¹³⁶ It generally follows that a patient innovation used to treat a specific orphan disease would result in a real world context of use. Thus, it would meet the “substantial utility” requirement.

As long as a patient innovator identifies a specific orphan disease the innovation is directed towards, the claimed subject matter would meet the utility requirement for patentability under 21 U.S.C. § 101.

4. § 101 – SUBJECT MATTER ELIGIBILITY

Patent claims must be directed towards patentable subject matter.¹³⁷ The relevant statutory provision states: “Whoever invents or discovers any new and useful process, machine, manufacture, or composition of matter, or any new and useful improvement thereof, may obtain a patent therefor, subject to the conditions and requirements of this title.”¹³⁸ The terms “process,” “machine,” “manufacture,” and “composition of matter” limit

¹³¹ Oliveira, *supra* note 41, at 7.

¹³² *In re Fisher*, 421 F.3d 1365, 1371 (Fed. Cir. 2005).

¹³³ *Utility Requirement*, *supra* note 127.

¹³⁴ *Fisher*, 421 F.3d at 1371.

¹³⁵ *Utility Requirement*, *supra* note 127.

¹³⁶ *Id.*

¹³⁷ 35 U.S.C. § 101 (2017).

¹³⁸ *Id.*

the subject matter that is eligible for patenting. Courts have also limited the subject matter that is eligible for patenting. Specifically, in *Diamond v. Chakrabarty*, the Supreme Court reiterated that the “laws of nature, physical phenomena, and abstract ideas have been held not patentable.”¹³⁹ These are all examples of a judicial exception. Patient innovations that solely recite these exceptions will not be patentable. If a claim is directed to a judicial exception, the claim needs to amount to significantly more than the exception itself.¹⁴⁰ This investigation is specific to the facts of each innovation, so a blanket statement of patentability of patient innovations cannot be made.

V. RECOMMENDATIONS

Current efforts to foster research on orphan diseases should be supplemented by solutions to encourage development of non-pharmaceutical treatments. Additionally, sharing and commercialization of innovations made by the user innovation community should be bolstered. Patient innovation exists within the orphan disease community. However, patients need to be incentivized to share, or diffuse, their innovations to maximize impact of user innovation solutions.

The current focus on pharmaceutical treatments disregards non-drug innovations that are cheaper and just as effective at treating the disease.¹⁴¹ Currently, the Orphan Drug Act provides grants to orphan drug developers, but it doesn’t provide funding for patient innovators outside of the pharmaceutical space.¹⁴² Orphan Drug Act and patent eligibility for drugs should, however, be maintained as patient innovators are unlikely to develop drugs on their own due to their complicated, time intensive, and expensive nature.

To shift some focus to non-pharmaceutical treatment innovation, contests available only to user innovators could be created. Due to the small market size, the availability of a reward for innovation of an orphan disease treatment is high-risk. Thus, contests must take away some risk from inventors with push, rather than pull, incentives.¹⁴³ Assuming users

¹³⁹ *Diamond v. Chakrabarty*, 447 U.S. 303, 309 (1980).

¹⁴⁰ See *Alice Corp. v. CLS Bank Int’l*, 573 U.S. 208, 221 (2014).

¹⁴¹ For example, a Marfan syndrome patient developed “a textile mesh support for dilated heart aorta.” Oliveira, *supra* note 41, at 2. Additionally, a caretaker of a child with Angelman syndrome discovered that the child’s physical abilities were improved by filling a room with floating balloons where the child jumped and reached for them for extended periods of time. *Id.* at 4.

¹⁴² 21 U.S.C. § 360 (2017).

¹⁴³ Pull incentives focus on revenue, while push incentives focus on cost. Christopher-Paul Milne & Joyce Tait, *Evolution Along the Government—Governance Continuum: FDA’s Orphan Products and Fast Track Programs as Exemplars of “What Works” for Innovation and Regulation*, 64 FOOD & DRUG

are innovating in an area with little or no patent eligibility, contests would provide patient innovators with financial incentives to innovate. If the innovation is chosen, the costs associated with developing that innovation will be recouped by the prize of the contest. The prize system could be run by government agencies, medical research centers, or private foundations. Although this is not a universal solution, as many innovations will not claim a prize, it will serve to supplement the programs already available to orphan disease research.

To ensure widespread access to newly developed treatments, sharing by user innovators must be encouraged. Development and diffusion are incentivized when the cost of sharing is low.¹⁴⁴ The cost of sharing within the traditional patent system is high due to application fees and upkeep fees. One way to reduce costs of sharing is an exchange platform for patient and caregiver interactions.¹⁴⁵ Such a platform has been developed to inventory developed solutions.¹⁴⁶ Additionally, infrastructure supporting exchange and communication between patients and doctors would decrease the cost of sharing. Increased communication between patients and their long-term doctors has previously achieved success in discovering treatment in other disease sectors.¹⁴⁷ A platform for exchange of solutions between patients and doctors would certainly lower the cost of sharing solutions, but the communication between doctors and patients can also extend to the testing of those solutions.

Primary care physicians and their patients diagnosed with an orphan disease can run their own trials on pharmaceutical and non-pharmaceutical treatments. Although primary care physicians and patients may be viewed as not sophisticated enough to run trials, they are the ideal candidates because “they [have] been managing the disease for years.”¹⁴⁸

Infrastructure platforms can be fostered with targeted grants for the creation of orphan disease treatment databases. The grants could go to intermediary organizations, such as NIH or NSF, to universities, or

L.J. 733, 739 (2009). For example, push incentive programs “result in R&D cost sharing or subsidies” and pull incentive programs “create incentives for private sector engagement.” *Id.*

¹⁴⁴ Canhão, *supra* note 52, at 33.

¹⁴⁵ *Id.*

¹⁴⁶ See PATIENT INNOVATION, <https://patient-innovation.com/> [<https://perma.cc/859F-XJ88>].

¹⁴⁷ See STEVEN EPSTEIN, IMPURE SCIENCE: AIDS, ACTIVISM, AND THE POLITICS OF KNOWLEDGE 216–17 (1996). Primary care physicians and people with AIDS circumvented the traditional clinical trial process for drugs by designing their own clinical trials. *Id.* at 216. “The idea was that physicians would distribute drugs, monitor patients, and collect data as an integral part of their regular clinical work with patients.” *Id.* at 216–17. Patients participated equally in decision making on critical aspects of trials. *Id.* at 217. This idea was also used in the National Cancer Institute’s community-based research effort. *Id.*

¹⁴⁸ *Id.*

established communities made up of patients, caregivers, and doctors. Instead of targeting grants toward a specific disease, the grants should be targeted towards broad infrastructure development for all orphan diseases. These infrastructure platforms will only succeed if “patients . . . who want to share information or organize a critique of medicine [can] seek out like-minded individuals and find points of commonality with them.”¹⁴⁹ To increase the success of these infrastructure platforms, the platform should be widely promulgated across orphan disease communities, including patients, caregivers, and their doctors, to increase the database of users.

¹⁴⁹ *Id.* at 229.

