日本ライソゾーム病患者家族会協議会
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発言内容の要約

（ドラグラグの経験から学んだこと）

1.　小児希少疾患治療薬導入の現状と課題

・ライソゾーム病は 31 種類あり、先天性の疾患で多くは重篤で患者数は非常に少ない。（数人から数百人）
・2005 年にムコ多糖症Ⅵ型治療薬のドラッグラグを経験し、個人輸入で薬を入手。
　この問題は国会でも議論された。
・小児希少疾患の治療薬導入は非常に困難。
・米国では 20 社が治療薬を開発しているのに対し、日本の企業は 1 社のみ。
・米国と日本との GDP の比較からも、両国間の開発状況の差は顕著。

2.　研究・治療薬開発の取り組み

・国内外の開発および研究情報を調査し、新規研究を依頼するとともに、大学での研究を支援。
・新薬の国内開発、導入を積極的に支援
・基金を設立し、医師や理学部の研究者への寄付を行い、基礎研究を支援。

3.　新規プロジェクトと支援の必要性

・海外のシーズと日本の研究者とのマッチングをサポート。
・患者数が数人の疾患は採算性の面から製薬企業の協力が得にくい状況にある。
・産官学に患者の意見を加えた産官学患の連携の重要性。
・国内に数人の治療薬開発は研究者の協力が得にくい。

4.　患者家族の声が届きにくい現状

・ライソゾーム病は進行性の疾患であり、若くしてお子様が亡くなることも少なくない。
・介護などによる生活の制限から患者家族が活動を行うことが難しく、国に声を届けるのが困難な状況。
・この病気は時間との戦いなので、早期の治療薬の開発が必要である。

5.　政府の支援の必要性

・新生児マススクリーニングやゲノム解析などによって早期診断と早期治療が必要。
・市場原理に基づく医薬品開発が難しいため、政府の全面的な支援が必要。
・少ない患者の疾患だからこそ、政府の支援がなければ取り残されてしまう現実がある。

子供たちが一人も取り残されることのないよう、国を挙げての支援を強くお願い申し上げます。

Provisional Translation

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Executive Director Takeyuki Akiyama

Summary of Remarks
(Lessons Learned from the Drug Lag Experience)

1. Current Status and Challenges of Introducing Treatments for Pediatric Rare Diseases:

- There are 31 types of lysosomal diseases, congenital in nature, most of which are severe and have very few patients (ranging from a few patients to a few hundred).
- In 2005, we experienced a drug lag with the treatment for mucopolysaccharidosis type VI and had to obtain the medication through personal importation. This issue was also discussed in the National Diet of Japan.
- Introducing treatments for pediatric rare diseases is highly challenging.
- While 20 companies in the United States are developing treatments for these diseases, only one company in Japan is doing so.
- The disparity in the development status between the two countries is evident, even when considering their GDPs.

2. Efforts in Research and Drug Development:

- Investigate research and development information domestically and internationally, request new research, and support research at universities.
- Actively support the domestic development and introduction of new drugs.
- Establish a fund and make donations to medical doctors and researchers to support basic research.

3. Need for New Projects and Support:

- Support the matching of overseas seeds with Japanese researchers.
- Diseases with only a few patients face difficulty in getting cooperation from pharmaceutical companies due to profitability concerns.
- It is important that patient’s voice should be involved in industry-academia-government collaboration.
- The development of treatments for these diseases with only a few patients in Japan faces challenges in gaining researcher cooperation.

4. The Voices of Patients' Families are Hard to Reach:

- Lysosomal diseases are progressive, and it is not uncommon for children to pass away at a young age.

- Due to lifestyle restrictions from caregiving, it is difficult for patients' families to engage in activities and make their voices heard by the government.
- Early development of treatments is crucial because these diseases are a time-sensitive matter.

5. The Need for Government Support:

- Early diagnosis and early treatment are necessary through newborn mass screening and genome analysis.
- Comprehensive government support is essential due to the difficulty of drug discovery and development based on market principles.
- Patients suffering from rare disease will be left behind without government support. This is the reality.

We sincerely request the government's support to ensure that no child is left behind.