# The IOPD Information Pack Support for families with children with Infantile Onset Pompe Disease

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# Summary

Following an approach to Sanofi Genzyme from a Clinical Nurse Specialist at Great Ormond Street Hospital (GOSH), a significant unmet need was identified. Parents of children with infantile onset Pompe disease (IOPD) required support to gain access to mainstream school education. These parents rely on their specialist nurses to provide advice on a wide range of subjects. Discussions around enabling their children to go to school, and have their special needs addressed while in education, were beginning to take up a significant proportion of precious consultation time.

Through a truly collaborative approach, an innovative information pack was developed for parents of children with IOPD to support them in selecting an appropriate school and in having vital conversations about their children's needs. The pack was also intended to support the schools, raising their knowledge level about IOPD and helping them to provide the support and resources that would help children with IOPD get the most out of their education.

# Results

**Objective 1** Equipping parents:







This poster outlines the process through which the specific challenges facing parents were identified and how the pack's content was developed to provide tangible solutions. The results detailed herein illustrate the significant contribution that the information pack has made to the quality of life of children with IOPD and their families. It is hoped that this project will serve as an example of best practice to inspire future collaborations that address unmet needs in rare diseases.

## Introduction

Pompe disease is a very rare genetic disorder, which can affect infants, children and adults. It impacts muscle function with consequences for breathing, eating, sleeping, mobility and cardiovascular function. The infantile onset form (in children diagnosed under one year of age) is extremely rare and there are only eight families affected in the UK today.

Historically, most babies with IOPD died before their first birthday from cardiac/respiratory failure. With the introduction of enzyme replacement therapy, the natural history of the disease can now be modified and children are reaching the age where they can go to school.

The benefits of school attendance extend beyond learning the curriculum. It's where children build relationships, make friends and (if it's done correctly) have fun. It is a big step for children and their families, but it can feel even more stressful when the child has IOPD.

All families in the UK (8/8) of school-age children with a diagnosis of IOPD received the pack

All these families fed back that the pack was helpful in having conversations with schools about their child's needs

All specialist centres in the UK who support families of children with IOPD as well as the AGSD-UK are committed to disseminate the pack to other families when their children reach school age, providing a lasting legacy for the pack

It was very, very full of information and had everything I needed in the pack. It is in depth and I read the pack to prepare for meetings with the school so I knew exactly what to expect and exactly what to ask. It had questions in there that I had not even thought of asking or considered."

Mother to a child aged 10

It's a really good tool. We didn't have this when we started our son at nursery but wish we had. It helps give us advice about caring for a child with Pompe as well as schools. This is a good thing to show parents that there is going to be a life in the future, with opportunities, hopes and dreams. As a parent of a child with Pompe this is exactly the kind of information we need. The more awareness there is the better and the more information the better." Family with a child aged 5

# **Aims and objectives**

#### There were two target audiences:

- 1. The eight families of school-age children with IOPD in the UK.
- 2. The resources were developed to support parents in their discussions and arrangements with schools. Therefore, the audience also included school head teachers, administrators and local education authorities.

#### The objectives were:

- 1. To equip parents with information and tools to promote full discussion of their child's needs with the school or education authority
- 2. To equip schools to address the specific needs of each of these children
- 3. To provide resources to healthcare professionals so that they could support families more efficiently and holistically



## **Objective 2** Equipping schools:

Schools now have a resource to improve their understanding of IOPD and its treatment and their ability to meet the needs of the children has improved

"A few weeks ago I visited a family with a child with IOPD. The mother felt the new support **FF** assistants were not heeding her son's requests to use the bathroom and there were, consequently, many incidents of incontinence. He had never had these problems before. I wrote to the school, offered to visit to talk about Pompe and also sent the IOPD school resource booklets. The mother said today that everything has changed and they are being much more involved in her son's needs and there are no more continence issues. She is sure it is due to the leaflets and now they understand Pompe better."

Jane Lewthwaite, Association for Glycogen Storage Disease (UK)

"Our school keeps saying my son does not need a health care plan but the information in the pack is helping me to make a case." Family with a child aged 9



### **Methods**

The project began with an approach to Sanofi Genzyme from a Clinical Nurse Specialist at Great Ormond Street Hospital (GOSH). Parents were relying on her to advise on how they could enable their children to go to school, and have their special needs addressed while in education.

There was literally no comprehensive source of support and advice for parents of children with IOPD about access to school education. Because there are so few children with IOPD in the UK, metabolic clinical nurse specialists spent considerable amounts of time supporting parents with children at this important life stage.

While parents are acutely aware of their own child's specific needs, they are often reluctant to let others care for their children. Meanwhile, schools cannot be expected to know where to start in putting the right support and staff training in place. The established UK patient group, the Association for Glycogen Storage Disease (AGSD-UK), did not have any specific resources for IOPD around this topic and welcomed the opportunity to collaborate to address this gap in support resources.

The strategic approach was to collaborate deeply with all the stakeholders (families, healthcare professionals and patient organisations) to develop the right content and present it in a way that met their needs.

- Understanding the audience: Pompe disease is part of a complex scientific area. Parents are highly educated on this disease and they become experts. Therefore it was important to use a writing style that was accessible, but not patronising. Schools would need more disease information than parents, set in the context of how they could support the needs of the child.
- **Providing the right content:** It was essential to cover content of relevance to the families and to the schools. The only way to ensure this was to not simply 'involve' these stakeholders, but purposely to have key stakeholders drive the content.
- The content development process: A core steering group was set up in September 2016, comprising specialist nurses, a dietitian, physiotherapist, speech and language therapist, occupational therapist, the patient organisation AGSD-UK and parents whose children had made the transition to school. They developed a content outline from which a medical communications agency wrote a comprehensive information pack with sections for parents and for schools. The pack would be disseminated to parents by metabolic clinical nurse specialists, and the AGSD UK as well as to schools via the parents.
- In addition to the core steering group, both other specialist treatment centres that treat children with IOPD in the UK reviewed and approved the complete pack and the AGSD-UK also reviewed and endorsed it.

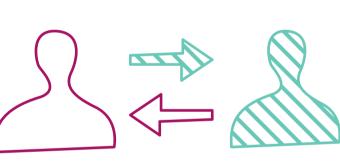
#### Pack contents: The pack was launched in September 2017.



The parent sections help families to understand their rights and discuss with their chosen school the changes and adaptations that might be needed to fully support the child's development. Materials include:

- Information to guide parents in the care of children with IOPD who are reaching school age
- Information on how to complete an Education, Health and Care (EHC) plan
- Checklist for use when visiting/selecting schools
- Information on the needs of the family, including emotional, physical and functional support







At GOSH alone the specialist nurse has saved "hours" of time that she can now devote to other clinical and support priorities

The pack therefore has improved conversations between the healthcare professional and the patient's family

In addition to these tangible outcomes, a market research survey with 12 specialist physicians conducted after the IOPD pack was launched revealed that the packs had improved their perspective on the reputation of Sanofi Genzyme

"A Pompe patient that was starting school - parents asked for advice on where to send them but there was no information available. I spoke to Sanofi Genzyme and they had a leaflet to signpost patients to an appropriate school/what to look for in a school." Specialist Nurse

#### Conclusions

Rare diseases often don't get the same recognition as other conditions in terms of pharmaceutical company investment 'beyond the product'. In Pompe disease, there are very few specialist treatment centres and very few families affected. So why would any pharmaceutical company reach out to them? Sanofi Genzyme believed that even if one person benefited, they were prepared to invest that time.

In some cases, pharmaceutical companies involve patients in commercial activities that the company wants to do. This project was completely the opposite. The target audiences were fully engaged in identifying the challenges and developing solutions. This was true collaboration led by a clear unmet need.

This project may only reach a small number of people, but for those it reaches it means the world. This pack means that each child with IOPD can have the best experiences no matter how long or short their life is. It means they can make friends, go on school trips, learn about the world and, for a short while, put their own stamp on it.

This project demonstrates:

- A gold-standard approach to real patient-centric communication and support
- The power of true collaboration between healthcare professionals, patients and the pharmaceutical industry
- A measurable benefit to patients in terms of inclusivity and access to education
- A measurable benefit to the precious time resources of the healthcare professionals who care for these patients
- The reputational benefits available

Although schools are not required to physically manage IOPD, they need to be aware of their role in reporting to the child's care team. The school sections of the pack were developed to help a school understand the impact IOPD has on a child and their family and how to support them:

- Background on Pompe disease and its management
- Checklist of requirements to ensure the environment is inclusive for children with IOPD
- Information on equipment and adaptations that should be made to support a child with IOPD

This approach should inspire other companies, not just in rare diseases, to collaborate more deeply with stakeholders and support patients' broader unmet needs.

for pharmaceutical companies that choose to push their role beyond the commercial aspects of provision of medicines

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Inclusive education for a child

Support

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