Look – a very good solution!

Yes, that’s a great idea

GAUCHER BEST PRACTICE

A broad collection of good practices from patients for patients

International Gaucher Alliance
Driven by passion for patients
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Gaucher organizations worldwide have a common voice in the International Gaucher Alliance (IGA), which is the umbrella organization of 48 national organizations. Treatment of Gaucher patients varies in the IGA member countries. As there are many examples of good practices in individual countries, the IGA would like to help patients improve their daily life and to minimise Gaucher Disease related stress and difficulties.

A working group comprising Thomas Biegler (Germany), Davor Duboka (Serbia), Gil Faran (Israel), Jasenka Wagner (Croatia) and Irena Žnidar (Slovenia) have collected the examples of good practices in this brochure. Our focus is on the daily life of Gaucher patients.

The examples were collected in 2015 through a questionnaire asking about the daily life of Gaucher patients. 131 patients from 22 countries have contributed to the brochure, which we encourage you to use, enjoy and learn from. Share it with your family, your doctor at the hospital or in other situations, where suggestions could be helpful.

We would be delighted if these good practices would help patients to improve their daily life with Gaucher Disease and would be happy if you send us your further thoughts, ideas and best practices so that we can share them with the community.
Routine Monitoring Tests

People with Gaucher Disease routinely go to hospital or to a medical center to have tests taken (e.g. blood tests, bone density, magnetic resonance imaging, ultrasound) – on this page please find examples of good experiences and good advice for use in the meeting with the doctor at the health care center or hospital.

“Take responsibility for yourself and for your check-ups
You should not wait for calls from the medical center or from the medical nurse regarding time for a check-up”

“A good spirit from your side and willingness to cooperate with the medical staff is a must”

“For children, the regular checkups are done during school holidays, so they are not absent from school”

“Be sure to ask for a written report after each routine check-up and make sure that you receive it within a reasonable time”

“You should request an explanation of treatment procedures and necessary check-ups”

“Ask for information material on treatment and procedures. Such material leads to a better understanding of the full treatment and a better compliance”

“Take responsibility for yourself and your family – be familiar with the treatment and the processes
Write down questions before you go to a consultation and make sure to ask questions”
“Ask your specialist to highlight abnormal findings during the routine check-ups. This could be followed by an update from the specialist over the phone according to his/her judgement.”

“Don’t waste your time while waiting, spend the waiting hours reading or playing”

“Check-ups work best when patient, doctor and other medical staff are flexible and medical examinations and dates are coordinated properly.”

“You get the best experience when the medical staff is flexible and committed and if the medical staff is available when you need it.”

“Line up in advance, this will reduce testing time, especially when many patients queue up for tests.”

“You get the best result when a specialist clinic familiar with Gaucher Disease conducts your routine check-ups.”

“The most optimal situation is when the whole check-up process with all tests takes place on the same day and at the same place.”

“After having received the results of your check-up ask your specialist whether you, if needed, can get another consultation or maybe an expert consultation by phone.”

“After having received the results of your check-up ask your specialist whether you, if needed, can get another consultation or maybe an expert consultation by phone.”
All Gaucher patients from my country travel on the same day to the capital where we all have all monitoring tests. This is also an opportunity to meet each other.
One specialist monitors all Gaucher patients (e.g. for bone density, ultrasound, magnetic resonance imaging).

I always go to the same specialist (e.g. radiologist) and to the same MRI/DXA machine, with the same setup, so that my test results may be compared from year to year.
Enzyme Replacement Therapy (ERT)

Enzyme replacement therapy works by replacing the missing or deficient enzyme (glucocerebrosidase). ERT is administered through intravenous infusions every 2 weeks, either at an infusion center or at home.

“I store the drug at home and take it in a cool bag to school, where I get the infusion at my school hospital”

“I now receive the medicine in the city where I live. This is much easier and it does no longer take several hours to go for infusions”

“Our doctors order the medicine and we just have to bring the prescription from the family doctor once a year”

“We store the medicine at home (it could be in a special refrigerator) and bring it in a cool bag to our family doctor, who gives the infusion”

“The hospital prescribes the medicine. The pharmacy prepares the drug and delivers it to the hospital where I receive the infusion”

“I receive medicine at the regional hospital or at the doctor’s office – this significantly improves my quality of life”

Infusions at the clinic or hospital

Enzyme Replacement Therapy (ERT)

Enzyme replacement therapy works by replacing the missing or deficient enzyme (glucocerebrosidase). ERT is administered through intravenous infusions every 2 weeks, either at an infusion center or at home.
“The hospital unit is flexible within its opening hours regarding time for infusion”

“I have the infusion every two weeks at the hospital (on Tuesdays)
If I need to I can come any other day of the week”

“I get the medication delivered when I arrive at the hospital department and I stay in a room where I mix the medicine and get ready
The nurse only inserts the needle/a cannula
I go to the hospital cafe until I’m done (I switch to salt water and get flushed) then I get “disconnected” and I’m ready to go home”
Infusions at the clinic or hospital

“We can choose when we are able to/want to go It is also possible to arrange treatment at the local hospital if you live too far from the main hospital”

“I usually help with the infusion, but I find it nice to have someone around who can help me if something unforeseen happens”

“It is very difficult to find my son’s veins, when he shall have his infusion but the nurse is very patient with him and with the process”

“I meet other patients during infusion We share experience living with Gaucher Disease and have fun together”

“The nurse comes to every place in the country even if I am away from home and helps me with the ERT”

“The nurse at the health center arranges the time for infusion I have an option to change the time if it is not convenient”

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Home treatment

Gaucher patients have 3 options of treatment at home:

By patients on their own

I received a good training by the nurse and I feel confident having the infusion even when I am home alone.

By family/friends

My father prepares everything for the infusion and he inserts a needle into my vein.

By specialized nurse

I receive ERT from a homecare company.
"The nurse who comes to my home is very nice, warm and caring. She even gives my daughters small rewards for each treatment. The atmosphere is very pleasant and relaxed."

"In connection with annual follow-up of children, a special nurse checks the parent's skills regarding home treatment (e.g., mixing of drug, using venflon and canula and giving the infusions). I have good guidelines on how to handle the infusions."

"I take my infusions at home: On the day of the infusion, a courier picks up the medicine at the hospital pharmacy. The medicine is in a sealed box kept at a cool temperature. The courier delivers the medicine at lunchtime and a nurse opens the box at the time of the infusion."

"The nurse comes home to the family. This makes the entire process painless and even enjoyable."

"I plan my infusions every other week to fit in with the family life. I take the infusions in the evening while watching TV."

"An oncologist prescribes the drug. I receive home treatment by the help of a health care provider. They organize it and help me get the infusion at home."

"Make other people aware that you take infusions at home: I have asked a neighbor – who is a nurse to help."

"The health care provider delivers the materials to my home and the nurse delivers the drug."
“I pick up the drug at the local pharmacist in my home-town just before having ERT at home. I bring the drug home in a cool bag and store it in a kitchen refrigerator (I check that the temperature is 2-8 °C) until I have my home infusion. My Gaucher doctor gives me the receipt which is valid for 10 months.”

“We order medicine at the specialist nurse and pick medicine up at the hospital for 4-6 months. The medicine is stored in a refrigerator at home.”

“I collect the medicine and remedies at the hospital at the routine checks twice a year. I store the medicine in a special refrigerator, which I have received from the patient association (since I store a lot of medicine for 6 months) it would take up too much space in our own refrigerator.”
Vacation

Can you travel with Gaucher Disease? Check this out – with help of your national Gaucher patient organization.

It is possible for me to receive the infusion at another hospital in the country

I bring medicine and equipment on longer trips as agreed with my doctor
“I bring a ‘to whom it may concern letter’, from the doctor
The letter describes the medicine and the remedies
and gives a brief description of Gaucher Disease
as well as contact information to the doctor”

“I am flexible when going on a longer vacation – I receive a higher infusion dose when I am back”

“When I went on vacation, I just brought all the supplies and the drug
When I took the infusion I hung the bag on a coat hanger or on a safety pin for the curtain”

“If I go for longer vacation, I can skip 1-2 infusions”

“I have been both camping and at flights with medicine for ERT- everything is possible”

“If I go to vacation for 3 weeks, I have my ERT (home therapy) just before I go and soon after I am back home”

“I was studying/working abroad – I received the ERT at the hospital there (organized by the help of nurse, doctor, patient association, pharmaceutical company)”
Many people with Gaucher Disease prefer to take their infusions at home, as this gives more flexibility and an opportunity to make it part of the daily life. This demands that all equipment is at home, and some creativity if you for example do not have an intravenous stand and have to use other ways to hang up the infusion.

“I use only good quality plaster”

“I use a small pump
The pump enables me to move around
With the pump I can actually do everything, even driving”

“I use an electronic pump – this makes the timing exact”

“A telescopic intravenous stand – designed for home treatment, does not remind you of the hospital
The weight is 2 kg, when folded it does not take much space (70 x 10 x 10 cm) and is easy to carry”
“When I am home alone, I use Y-system, that means I connect both bottles (with the drug and saline for flushing) to the system. When the first bottle (with the drug) is empty, I only open the flow of the second bottle. When I don’t use Y-system, I ask somebody to change the bottles for me, as this is quite difficult to be done by myself having metal needle in the vein.”

“I have no stand. I remove a photo from the wall and use a nail to hold the set.”

“I have a home-made hanger.”

“I always use 0.2 micron filter (it is recommended to be used in the instructions leaflet)”
For each infusion
I receive an already prepared kit which contains:

- infusion set
- 0.2 micron filter
- venipuncture set (e.g. butterfly or intravenous catheter)
- 0.9% NaCl bag (e.g. 100 ml or 250 ml)
- water for injection (n x 10 ml)
- needles (e.g. 18 G or 19 G)
- syringes (e.g. 5 ml or 10 ml)
- plaster
- plaster to fix the venipuncture set (e.g. transparent film dressing frame style)
- skin cleansing alcohol prep pads
- nonwoven swabs
- gloves
- garbage bag

“I use a flexible plastic needle, so that I can move my arm”

“I receive material for one year, including everything”

“The nurse brings everything I need”
Substrate Reduction Therapy (SRT)

Substrate Reduction Therapy works by reducing the amount of glucocerebroside in the body. This process decreases the work the body’s enzyme must do by giving it less glucocerebroside to break down. SRTs are oral medications, taken once or twice daily.

“I write start and stop dates on the medicine strips to help me remember to take the capsules. This way I have exact data on how many capsules I forget in one month and in a year.”

“In addition to the use of an app to remind you should have a reminding system (for instance when brushing your teeth).”

“I have an alarm in the mobile phone as a reminder that I shall take my capsules.”

“I use an app, which helps me prove that I have taken the capsules. I send the calendar to my e-mail to give me a quick overview of when I might have forgotten to take the capsules – this is important to tell the doctor.”
Other aspects of living with Gaucher Disease

Social aid for families with children with Gaucher Disease (50-60 EUR per month) until the child is 18

Disability pension during the months of school for a child at school

Seven extra days leave from work

Getting some kind of tax allowance

Invalidity pension

Severely handicapped passport, more leave per year, dismissal protection, using public transportation for free

Early retirement

Young patients (18-26) may receive social aid if they are not able to work

I have no special privileges but I try to work one day at home during the week. This helps me save energy

All children with genetic conditions have status as disabled, this means that the parents have extra 3 days off per year, they receive a special allowance of 100 EUR, and the child may use public transportation for free upon showing a certificate.
Parents have the opportunity to seek compensation for loss of income in connection with hospital visits and the possible need of being completely at home with the child (for some Type 3 children).

Social security grant for children (under 18 and on treatment without complications):
Disability benefits, tax benefits and discounts on property taxes, electricity, vehicle registration.

Extra free days – depends on the company you are working for, we can be classified as “handicapped person”.

Early retirement extra social funds, reimbursement of costs of travelling.

Social security grant for adults (above 18 and on treatment without complications):
Medical disability and Vocational rehabilitation benefit (one life time vocational training, including university) and the rent at students hostels.

At regular Gaucher meetings (e.g. on International Gaucher Day, or at national and regional meetings) I meet other patients, families, doctors, nurses.
We share experience and learn more about different aspects of Gaucher Disease.
We are also in touch by e-mail and social media (e.g. facebook).
There are lots of good friends in the Gaucher community.

Longer meetings (e.g. for two days) are better than one-day meetings, as the participants become more relaxed and open, especially when we discuss difficult or personal topics.
My contacts, notes etc.
My experience is amazing treatment. I developed a real friendship with my nurse and my doctor who treats me with devotion, sensitivity, and provides a listening ear and emotional support.