

*Look – a very  
good solution!*

*Yes, that's a  
great idea*

# GAUCHER **BEST** **PRACTICE**

**A broad collection of good practices from patients for patients**



# Contents

|                                                           |    |
|-----------------------------------------------------------|----|
| Routine Monitoring Tests.....                             | 4  |
| Enzyme Replacement Therapy (ERT) .....                    | 8  |
| Infusions at the clinic or hospital.....                  | 8  |
| Home treatment .....                                      | 11 |
| Storage .....                                             | 13 |
| Vacation .....                                            | 14 |
| Materials and equipment<br>for intravenous infusions..... | 16 |
| Substrate Reduction Therapy (SRT) .....                   | 19 |
| Other aspects of living<br>with Gaucher Disease.....      | 20 |
| My contacts, notes etc. ....                              | 22 |



Statements used in this brochure are those of patients and their family members, collected for the purpose of this publication and are their own opinion and experiences and not those of the IGA.

Published by IGA  
Year 2018  
Authors: Irena Žnidar, Thomas Biegler, Davor Duboka, Gil Faran, Jasenka Wagner  
Written by: Randi Krogsgaard  
Graphic design and illustrations: Michelle Farrington  
Language: English  
Format PDF on [www.gaucheralliance.org](http://www.gaucheralliance.org)  
ISBN: 978-1-5272-1963-2

BE  
WE  
THE  
G  
D  
A  
G

# Sharing experience of the daily life with Gaucher Disease

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.

DISEASE



# 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”*

*“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”*

*“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”*

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

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

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



*"Don't waste your time while waiting, spend the waiting hours reading or playing"*

*"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"*

*"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"*

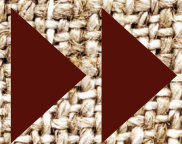
*"You get the best result when a specialist clinic familiar with Gaucher Disease conducts your routine check-ups"*

*"Line up in advance, this will reduce testing time, especially when many patients queue up for tests"*

*"You get the best experience when the medical staff is flexible and committed and if the medical staff is available when you need it"*

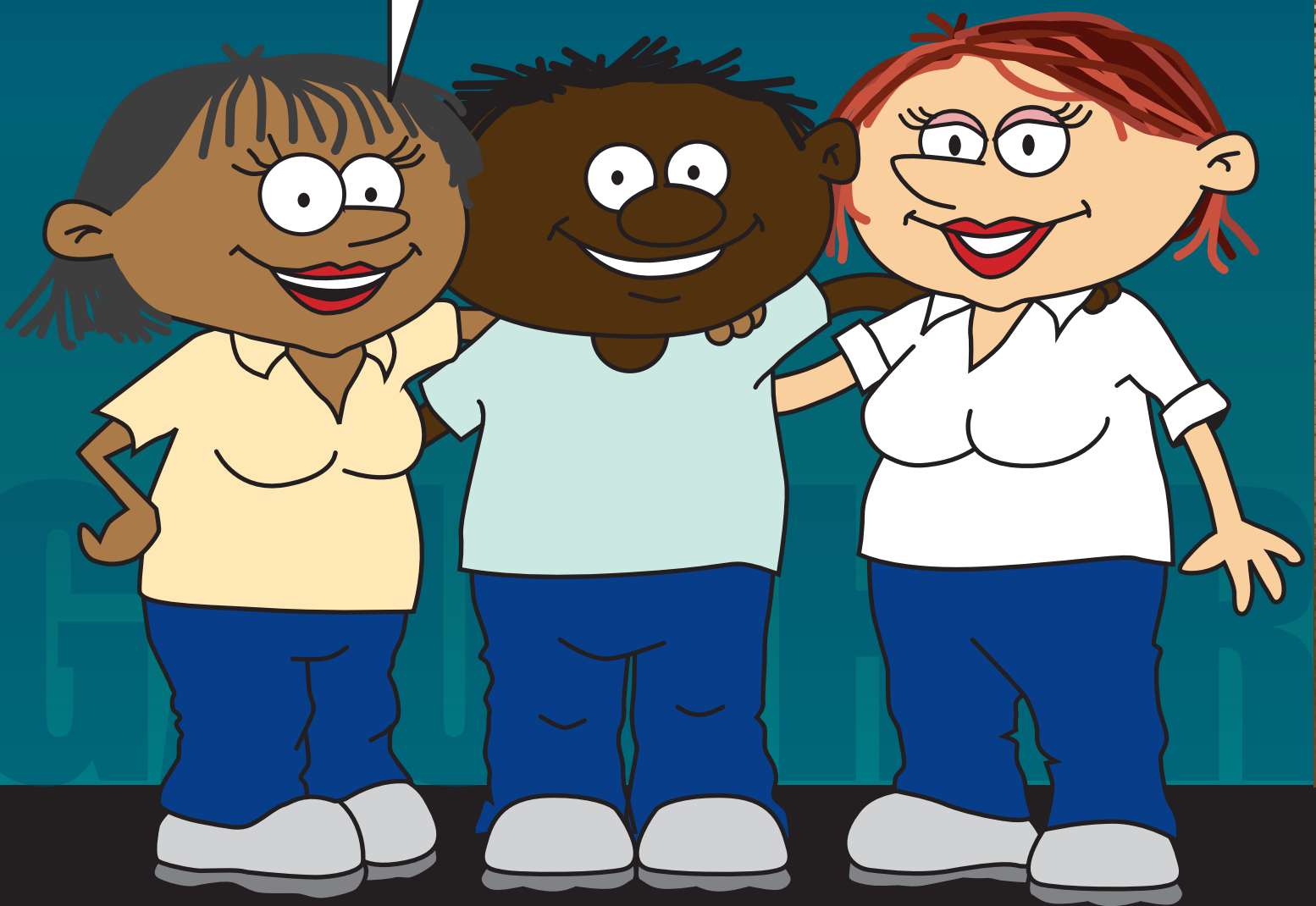
*"The most optimal situation is when the whole check-up process with all tests takes place on the same day and at the same place"*

*"Check-ups work best when patient, doctor and other medical staff are flexible and medical examinations and dates are coordinated properly"*





*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*

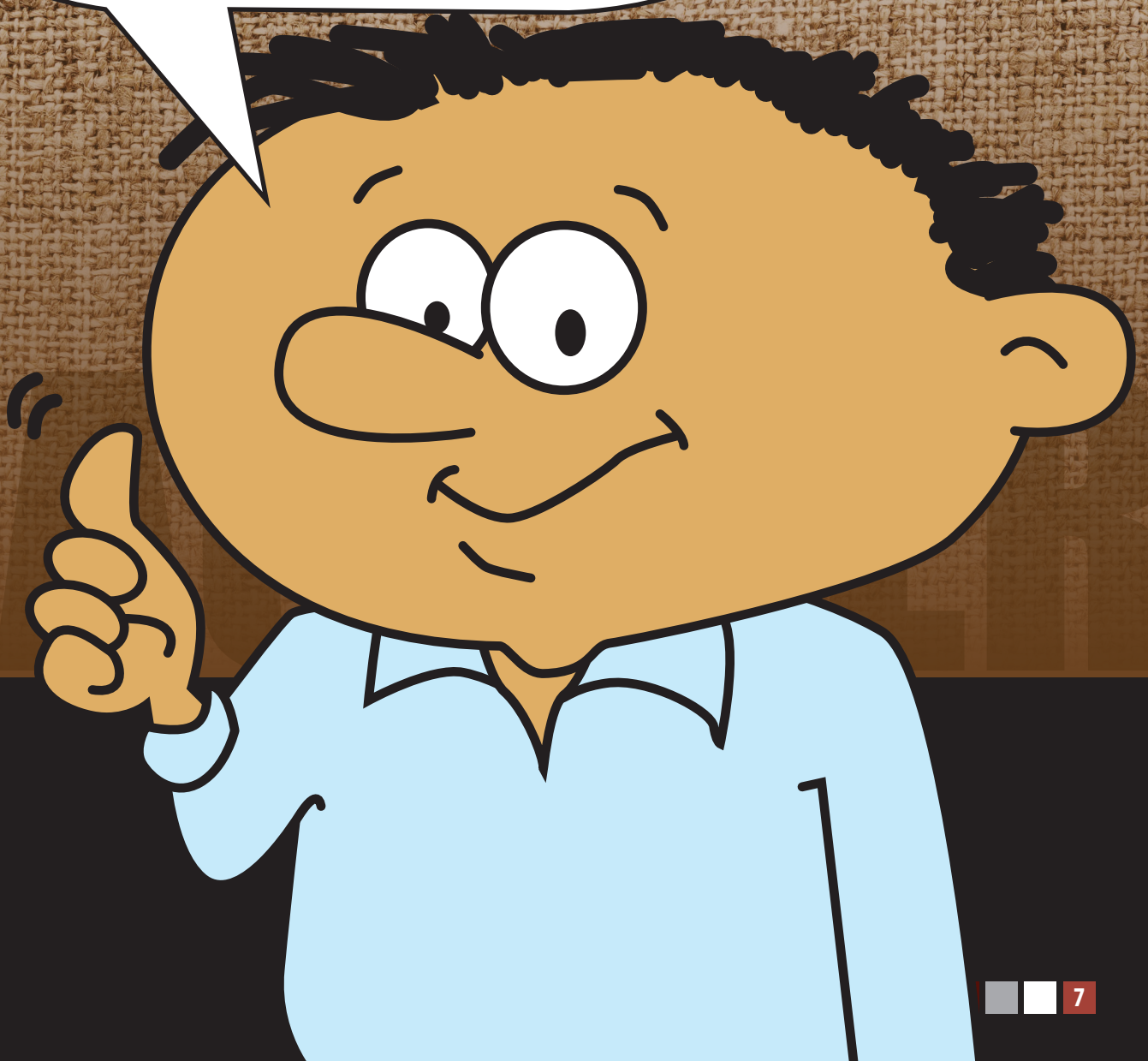




# One specialist monitors all Gaucher patients

(e.g. for bone density, ultrasound, magnetic resonance imaging).

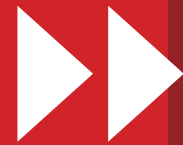
*I always go to the same specialist  
(e.g. radiologist)  
and to the same MRI/DXA machine,  
with the same setup,  
so that my test results  
may be compared from year to year*



# 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.

# Infusions at the clinic or hospital



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

*"The hospital prescribes the medicine*

*The pharmacy prepares the drug and delivers it to the hospital where I receive the infusion"*

*"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"*

*"I receive medicine at the regional hospital or at the doctor's office – this significantly improves my quality of life"*

*"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"*

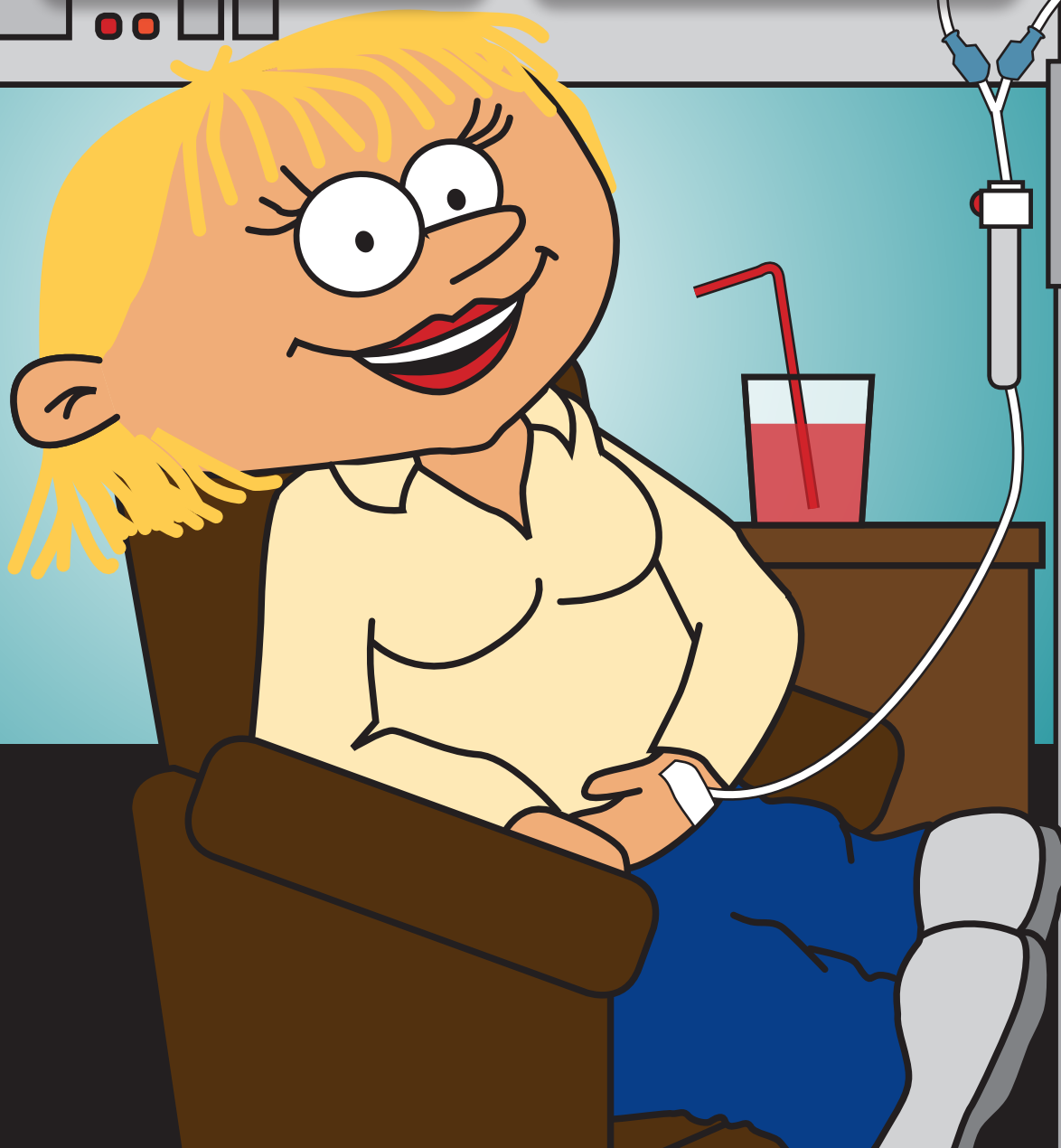


*"The hospital unit  
is flexible within  
its opening hours  
regarding  
time for infusion"*

*"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



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

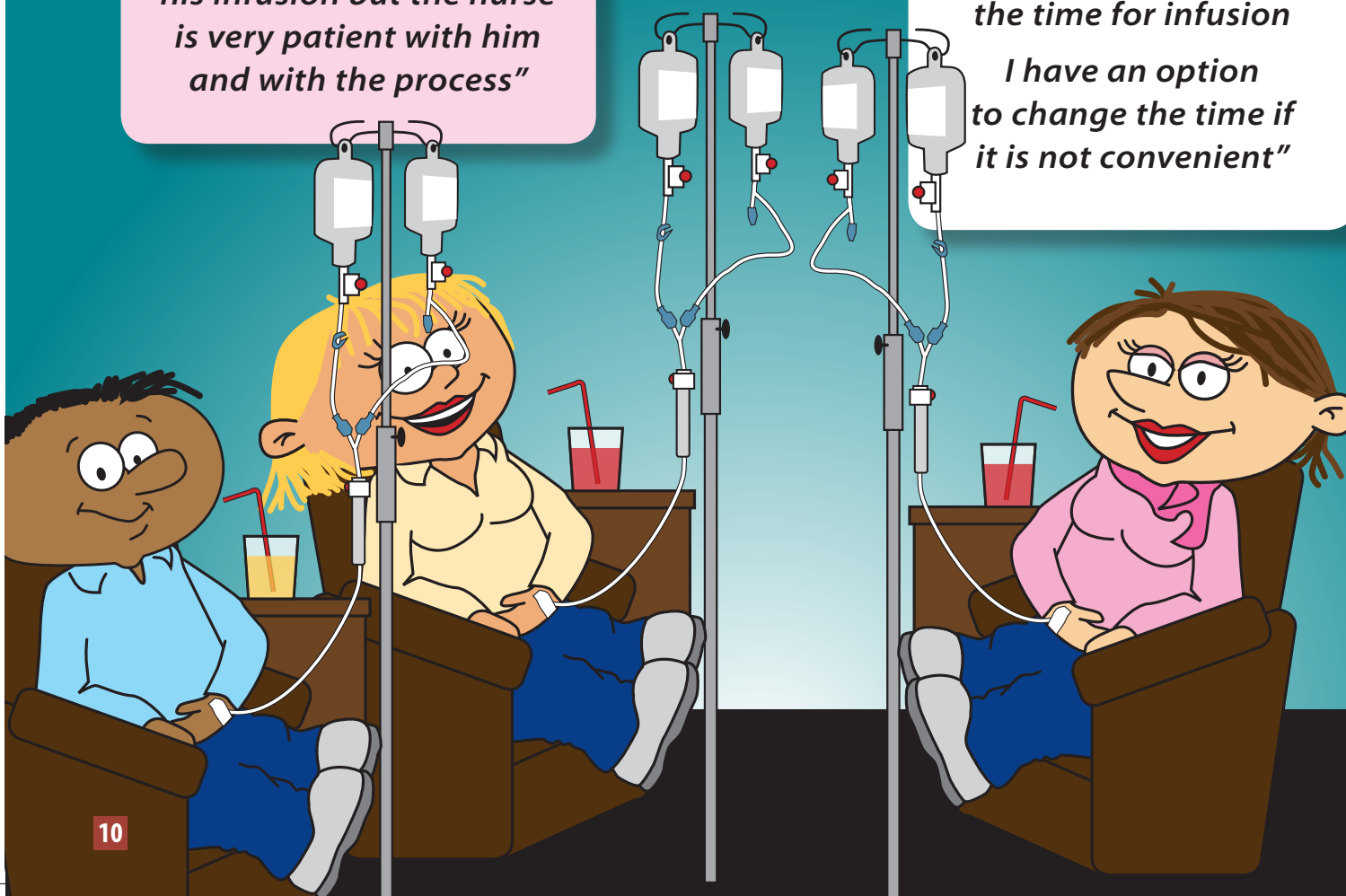
*"We can choose when we are able to/want to go  
It is also possible to arrange treatment at the local hospital if you live too far from the main hospital"*

*"I 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"*

*"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"*

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



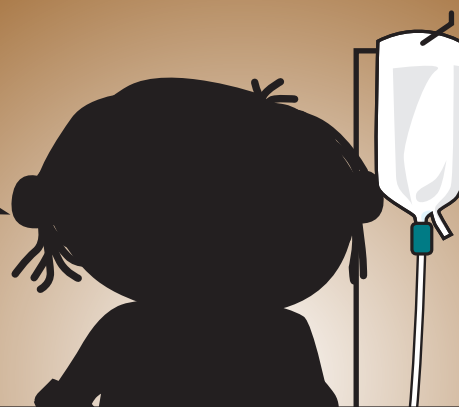


# Home treatment

Gaucher patients have 3 options of treatment at home:

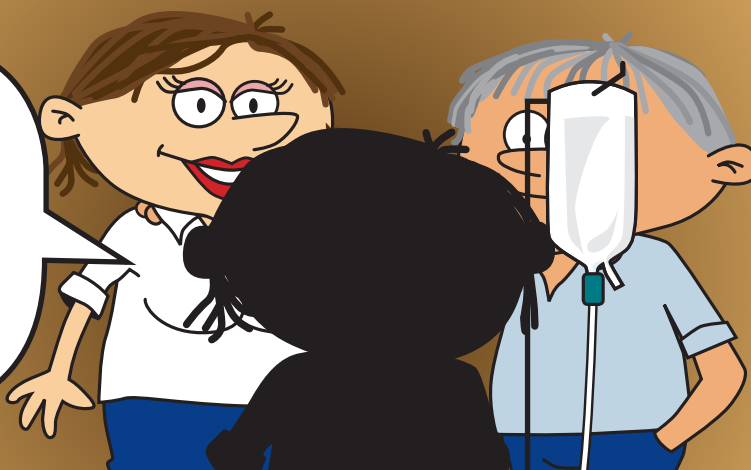
## By patients on their own

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



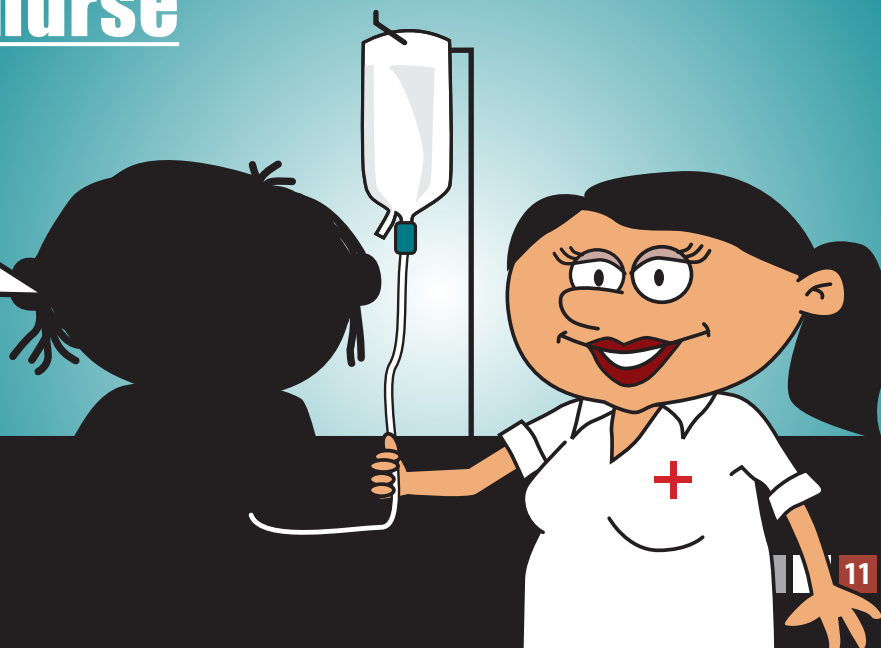
## By family/friends

*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"*



# Storage



*"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*

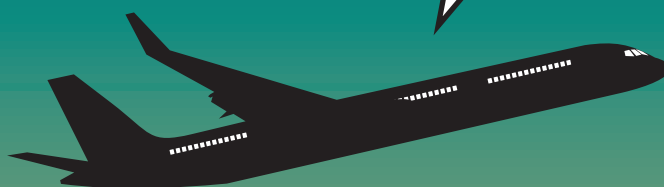
*I store the medicine in  
a special refrigerator,  
which I have received  
from the patient association  
(since I store a lot of medicine  
(for 6 months)  
it would take up too much space  
in our own refrigerator)"*

# 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*



# GAUCHER







*"If I go for longer vacation,  
I can skip 1-2 infusions"*

*"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 have been  
both camping and at flights  
with medicine  
for ERT- everything is possible"*

*"I am flexible  
when going on  
a longer vacation  
– I receive a higher  
infusion dose  
when I am back"*

To whom it may concern

*"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"*

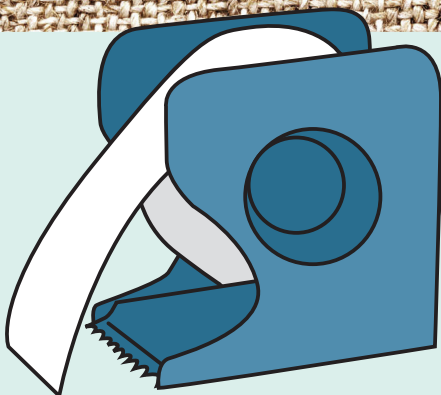
*"If I go to vacation for 3 weeks,  
I have my ERT (home therapy)  
just before I go  
and soon after I am back home"*

*"I was studying/working abroad  
– I received the ERT  
at the hospital there  
(organized by the help of nurse,  
doctor, patient association,  
pharmaceutical company)"*

# Materials and equipment for intravenous infusions

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"*



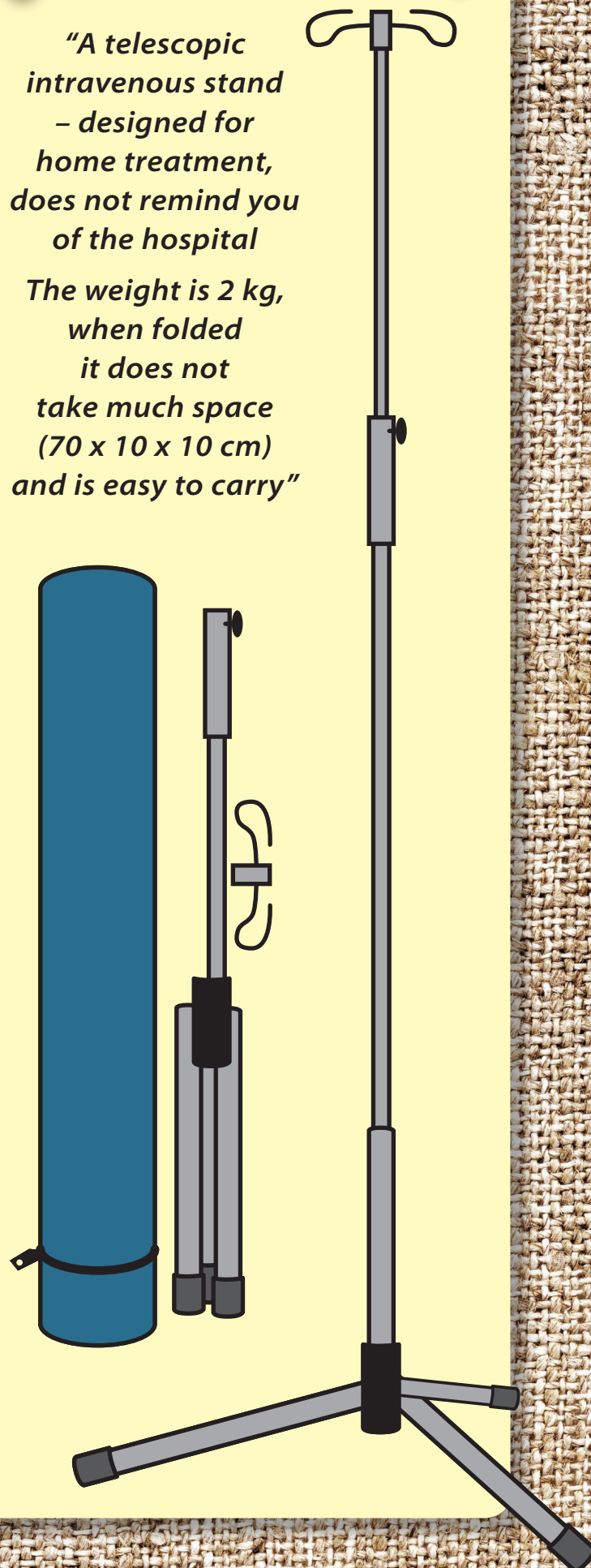
*"I use a small pump  
The pump enables me to move around  
With the pump I can actually do everything, even driving"*



*"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"*

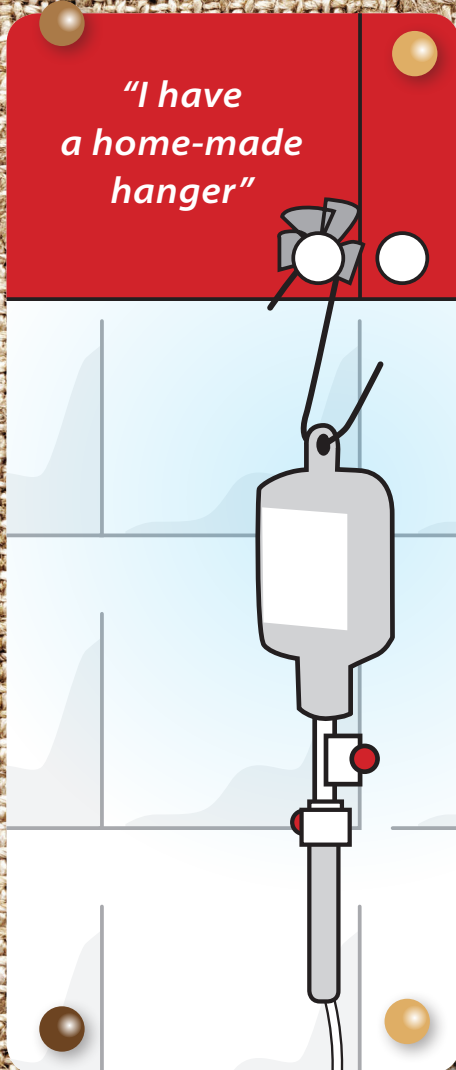
*The weight is 2 kg, when folded it does not take much space (70 x 10 x 10 cm) and is easy to carry"*





*"I have no stand  
I remove a photo  
from the wall  
and use a nail  
to hold the set"*

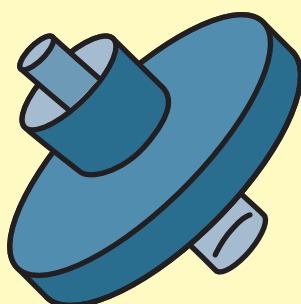
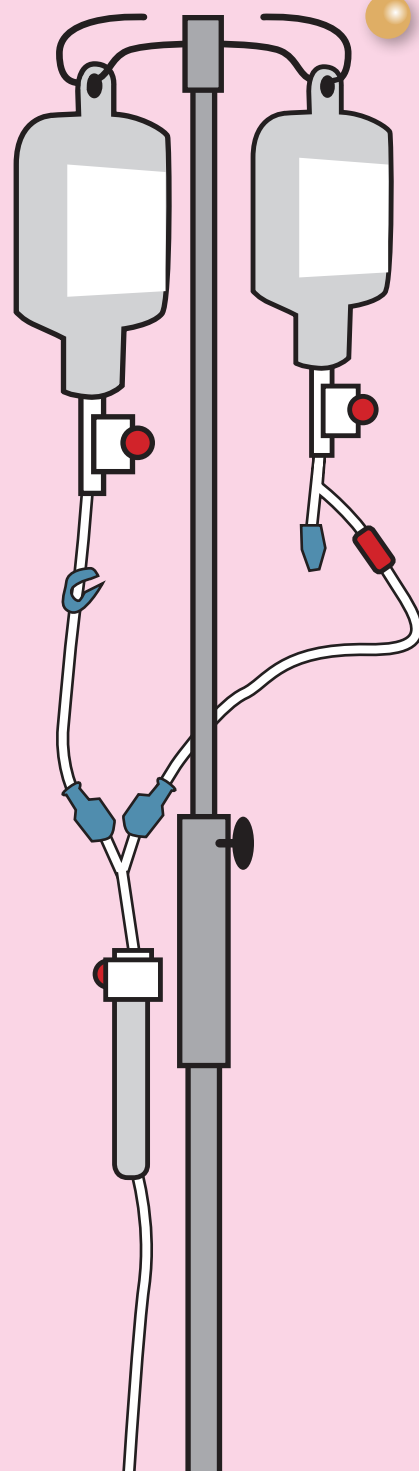
*"I have  
a home-made  
hanger"*



*"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*

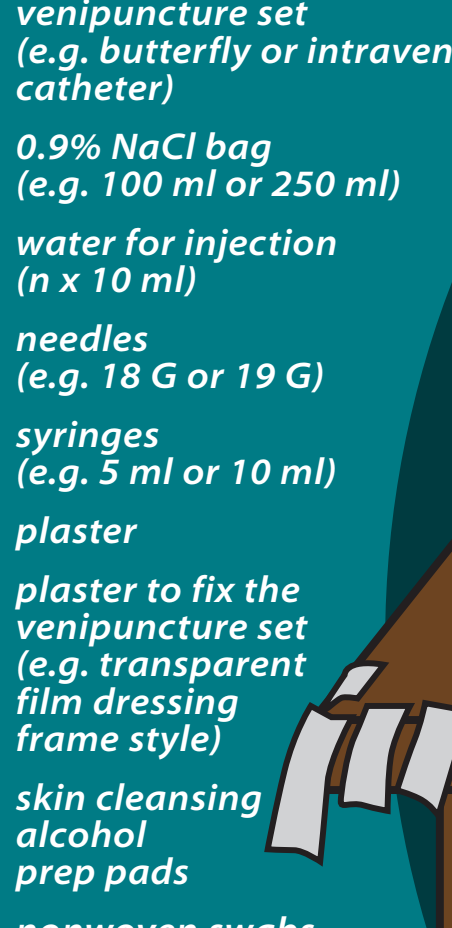
*When I don't use  
Y-system,  
I ask somebody to  
change the bottles  
for me, as this is  
quite difficult  
to be done by myself  
having metal needle  
in the vein"*

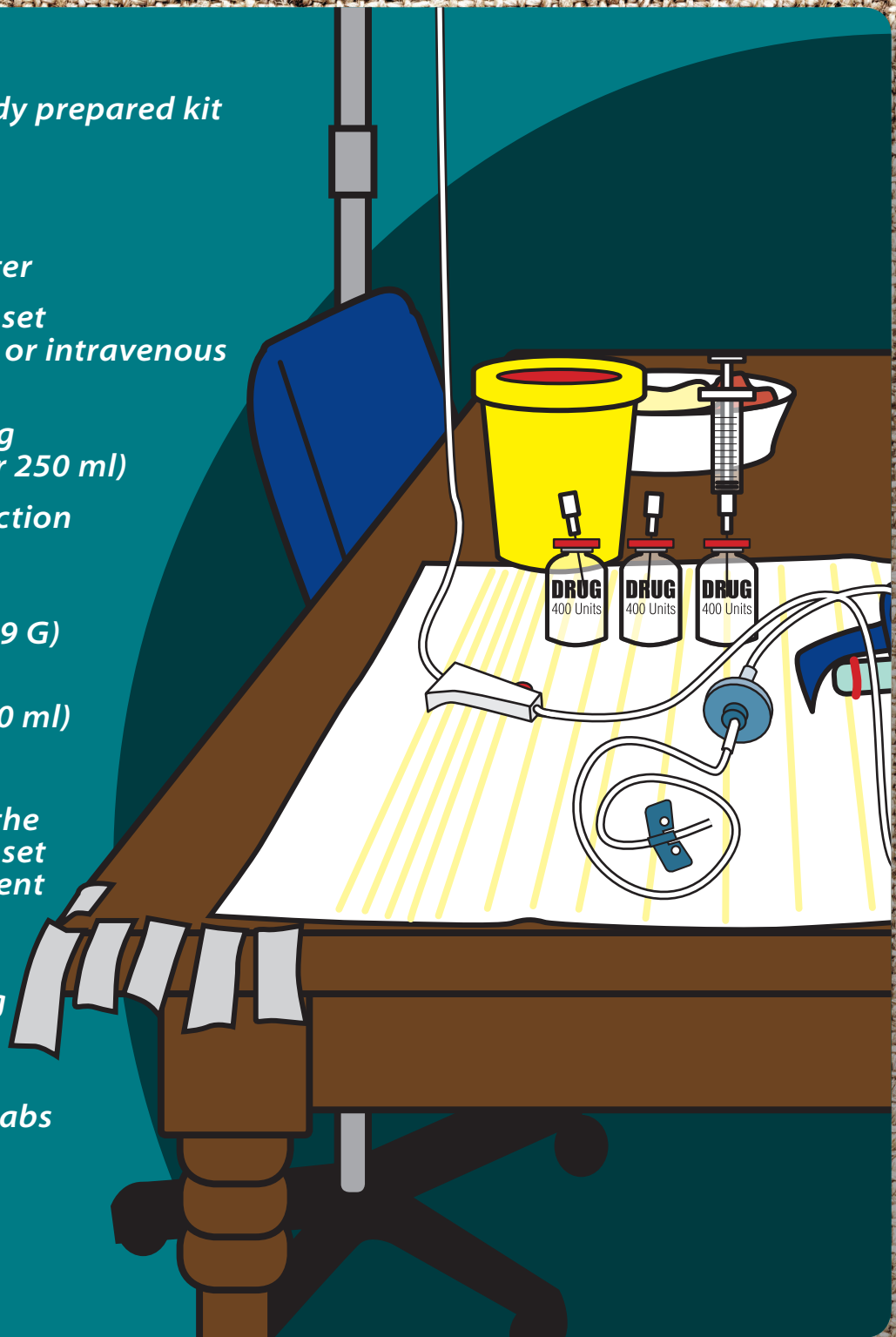


*"I always use 0.2 micron filter  
(it is recommended to be used  
in the instructions leaflet)"*



***For each infusion  
I receive an already prepared kit  
which contains:***

- 
- The illustration shows a person's arm with a venipuncture set inserted into the vein. A list of required items is displayed on the right side of the image, each preceded by a white square icon. The items are:
- 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 have an alarm in the mobile phone as a reminder that I shall take my capsules"*

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

*"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"*

*"I write start and stop dates on the medicine strips to help me remember take the capsules"*

*This way I have exact data on how many capsules I forget in one month and in a year"*



# Other aspects of living with Gaucher Disease

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

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

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

Seven extra days leave from work  
Social funds of 70% of minimum salary

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

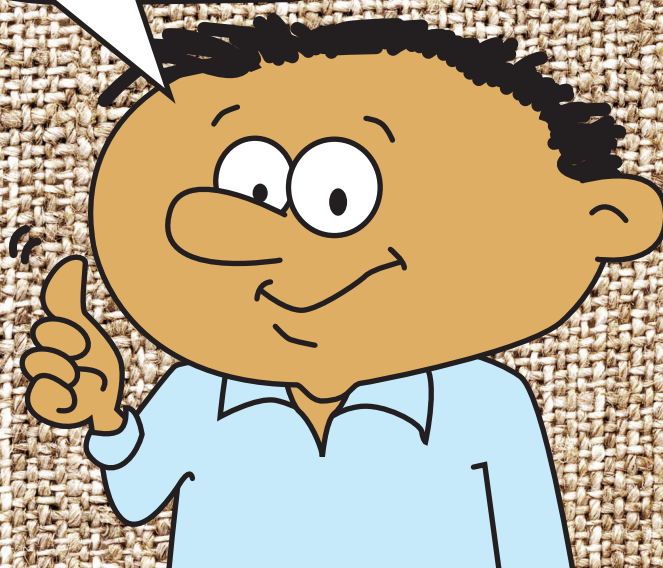
Getting some kind of tax allowance

Invalidity pension

Early retirement

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

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





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)

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 children (under 18 and on treatment without complications):

Disability benefits, tax benefits and discounts on property taxes, electricity, vehicle registration

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

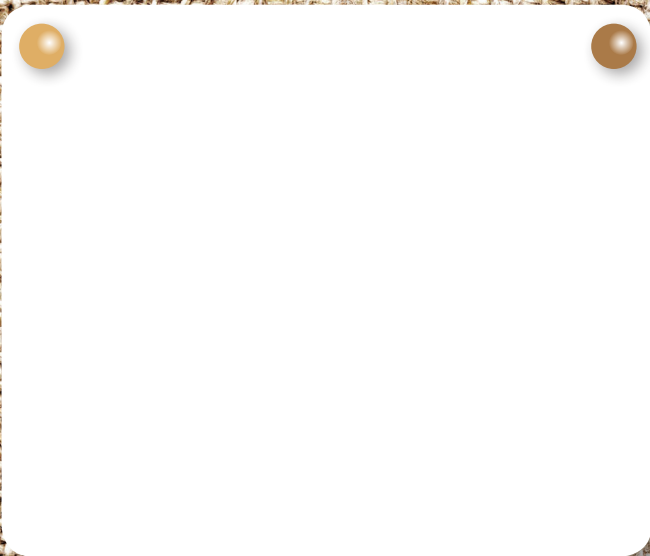
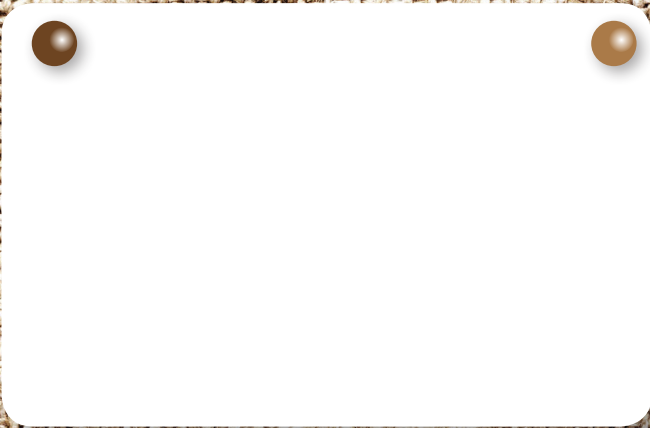
Longer meetings (e.g. for two days) are better than one-day meetings, as the participants become more relaxed and open, especially when we discuss difficult or personal topics





**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*

