

Carry your medical files with you

What is this about?

Thalidomiders may have a number of anatomical differences compared to others, both visible ones, but also those, which cannot be seen. The latter can have crucial importance when it comes to medical interventions and it is important that they be known to the treating health staff.

When students become doctors, they train on what is called “standard anatomical models”. While science does know that anatomic findings may differ a little bit from person to person, doctors generally do not know anything about certain anatomic features of thalidomiders. It will be up to the patient to tell them.

For example, if someone with „normal hands“ comes to the emergency room after sustaining a hand trauma, the x-rays will show the doctor if there is a fracture or not. The diagnosis is based on two observations:

1. because a fracture may show direct signs on the x-ray but
 2. because the doctor knows, what a non fractured „normal“ hand looks like on the x-ray.
- Based on that knowledge, he/she will be able to identify abnormalities.

Confronted with the x-ray of a thalidomide hand after hand trauma, the doctor will generally have no idea of what this hand would look like normally, meaning before the trauma

This lack of crucial information, bares the risk of misinterpretation of the x-rays and, as a consequence, decision of an inadequate treatment.

It is up to the patient to provide the doctor with the knowledge of what his/her hands looked like on x-ray prior to the trauma.

The approach is simple:

- Thalidomiders should have x-rays taken from arms and legs / shoulders / hips
- And save the files on data media like USB stick or CD which may be carried around as part of the every-day purse, or saved (password protected, maybe...) on an internet domain accessible for arising need.

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Suitable imaging :

The family doctor can advise on which images are best suitable for every case.

If no clear guidance can be obtained, I would suggest a conventional x-ray in frontal and a side view of each impaired limb. Feet and hands may require an additional 45° view.

Existing CAT and MRI scans may be saved to a USB stick or CD ROM, Simple saving as .jpeg is most likely to allow reading the files on any computer in the world. CAT scans and MRI are most helpful if the corresponding program for visualising the different layers of the images is also copied together with the images files.

Examples of typical thalidomide-associated anatomical alterations

1.) Congenital dislocation of the shoulder:

In the past, we observed several cases of thalidomiders with short arms who were admitted to an emergency room after an accident. Based on the examination of the shoulder of the patient, a shoulder dislocation was diagnosed and related treatment started. The treatment (closed reduction of the shoulder) lead to lead to catastrophic damage, because it is inadequate in the case of most thalidomide caused upper arm damage.

The shoulder articulation of thalidomide-caused short arms is completely different from a normal shoulder. This is apparent on external investigation and even more evident on x-rays.

Both the physical examination and x-rays indicate a shoulder dislocation. This is, however, a pre-existing condition (thalidomideers with severely affected upper extremities generally present severe congenital shoulder dislocation), to which the patient has adapted perfectly.

The shoulders should not be manipulated unless the patient complains about new pain and/or loss of mobility and shoulder function. Any attempt for closed reduction is likely to result in severe damage of the shoulder.

There is insufficient joint surface to allow for proper articulation; reduction may result in severe neurovascular trauma with no benefit for the patient.

Furthermore, it is unlikely that there is sufficient muscular fixation oft the shoulder with the (often dysplastic) rotator cuff and the joint capsule.

So, if a thalidomide victim with shortened arms is admitted to the emergency room, it is likely that signs of a severe shoulder dislocation will be diagnosed. In absence of other signs of trauma (bruises, abrasions) in the area of arms and shoulders, however, it has to be assumed that the shoulder dislocation is a pre-existing condition, which is normal for the patient.

Additional information must be obtained from the patient regarding his/her acute clinical symptoms (pain, loss of function, neurologic impairment) prior to any therapeutic attempt. If the patient is unconscious, postpone any treatment oft he shoulders until the patient can respond to your questions.

It is advisable for all affected thalidomides to carry a document which states the above condition and highlights the request for caution. At the end of this article I will make suggestions concerning such documents.

2.) Treatment of fractures is difficult, if not impossible since the anatomical specificities prior to the fracture is unknown.

While fractures of the long bones will most likely not go unnoticed and will presumably be interpreted correctly, it will be extremely difficult to identify fractures of thalidomide club hands or club feet due to overlapping bones in the x-ray. A good post-surgical result after osteosynthesis is unlikely if the pre-existing anatomical position of the bones is not well understood.

In some cases, a patient with a presumably broken hand was sent home by the doctor to wait for x-ray controls to indicate the bone healing on the edges of the fracture ends (build up of bone callus which can easily be identified in the x-ray). After 4 weeks, it was indeed easy to spot the site of the fracture, but it was too late for a surgical intervention. In these cases, pre-existing imagery like x-rays, CAT scans or MRI scans are mandatory to allow a comparison with the post-traumatic images and to guide the most adequate treatment.

3.) Altered or missing arterial vessels

The radial artery normally runs distally on the anterior part of the forearm. The artery winds laterally around the wrist and joins with corresponding branches of the ulnar artery in the deep and superficial palmar arch (anastomosis between two arteries), which ensures circulation of the hand.

Evolution is a master of efficiency, generally eradicating all superfluous tissue and it can be assumed that there is a very good reason for a redundant blood supply of the hand. The evolutionary idea behind an arterial anastomosis is, that in case one artery is injured, the other artery, connected by anastomosis will ensure the blood supply of the dependent region. Otherwise, injury to the radial artery would lead to loss of half the hand.

An absent radial artery is described as a (rare) variant in otherwise healthy people.

Patients with thalidomide embryopathy show a classic longitudinal damage pattern with a radial -> ulnar damage sequence, meaning that radial structures are damaged long before ulnar structures.

Initial hypothesis expected the radial artery to be intact in case of light structural arm damage caused by thalidomide. Recent MRI imaging of arms and hand of thalidomiders however seem to indicate that the radial artery can be affected even when there is only light structural arm damage. Several cases were described, where the only external stigma of a thalidomide malformation of the upper extremity consisted in a hypoplasia of the thenar (thumb) muscles, but where MRI scans showed complete aplasia of the radial artery. Apparently, blood vessel damage can exceed „orthopedic damage“ caused by thalidomide, resulting in loss of arterial structures in an otherwise only lightly affected limb.

The clinical relevance of these findings is due to the important role the radial artery has in medical diagnostic and therapeutic approaches.

- In coronary catheterisation, the radial artery is often used as point of entry instead of the femoral artery.
- Radial artery grafts have proven to be of superior value than venous grafts in aorto-coronar bypass surgery.

- Invasive blood pressure monitoring generally uses the radial artery as point of entry

In thalidomide patients (and due to the rare condition of a radial artery aplasia), an Allan test, that confirms sufficient blood supply of the hand by two arteries, has to precede any manipulation of the radial artery.

For a thalidomider who knows that he/she has no radial artery, it is advisable to carry a document in the personal papers stating this condition. In case of emergency, with this information, no time is lost finding the missing artery and an alternative arterial access (like e.g. femoral artery) is sought immediately.

If tests reveal that only one artery is functional, the manipulation of the artery ensuring blood supply to the hand must be restricted to life saving indications.

4.) Chest surgery in patients with tetra amelia.

While thalidomide also affected inner organs, the dysplasia of extremities was the most striking feature. Tetra-dysmelia and terta-amelia present the most extensive damage in thalidomidiers and involve severely deformed or missing arms and legs.

Persons affected by this condition learned as children to move along without the help of their extremities. Human will to locomotion is not hampered by missing arms and legs and persons with such extensive physical damage learned to move around by rolling or hobbling or by motion similar to that of a caterpillar.

This implies using both the thorax and abdomen for locomotion and needs to be considered in case of certain diagnostic or therapeutic approaches and with regard to the rehabilitation of the patient in order to regain the pre-existing level of autonomy.

Open heart surgery involving sternotomy will result in weeks of severe handicap and near immobility – thus helplessness - for any patient who relies on a stable thorax for locomotion.

In a situation, where endoscopic approach (PCI) or bypass surgery (ACVB) are adequate alternatives with respect to the coronary situation, the least invasive procedure (endoscopy) should be given preference in order to spare the patient unnecessary suffering from sternotomy.

In case of an indication for bypass surgery a thoracoscopic approach might lead to equally good results as open heart revascularisation but would spare the patient weeks and months of immobilisation after sternotomy.

Furthermore, due to the deformation of the legs, limited adequate vascular material for grafts has to be expected. A graft of the arteria mammaia may have to be considered as an alternative option.

Wording suggestions for your personal documents:

While there is a wide range of possible thalidomide originated defects, I am giving examples for the most common issues.

Every person should choose, edit, add and modify according to their own anatomical situation and personal needs and concerns.

It should be remembered that any therapeutic decision has to be the result of an understanding and agreement between doctor and patient. Informed consent for an intervention means that the patient be provided adequate information and “consents” (agrees) to the proposed intervention. Below wording suggestions may be helpful in case a patient is unconscious or too unwell to discuss.

Important documents should be carried in the every-day-purse in form of a Data CD, USB stick or printed media.

Suggestions in several languages

1.) Congenital shoulder dislocation

(GB)

The patient has a thalidomide caused embryopathy with **congenital dislocation** of both shoulders. The patient is well adapted to this condition. For detail, please refer to digital x-rays images included in the file. If current x-ray shows no alteration from the x-rays provided on file and if patient does not complain about acute shoulder problems do not attempt reduction the dislocation. The condition you see is normal for the patient. There is insufficient joint surface to allow for proper articulation; reduction may result in severe neurovascular trauma with no benefit for the pat.

(German)

Der Patient ist contergangeschädigt. Der Pat. weist eine beidseitige **congenitale Schulterluxation** auf. Ist an diesen anatomischen Status bestens adaptiert. Röntgenaufnahmen liegen als Bilddatei mit anbei. Sollten die aktuellen Röntgenaufnahmen einen identischen Status zu den vorbestehenden Aufnahmen auf diesem Speichermedium aufweisen und sind keine aktuellen Beschwerden im Bereich der Schultergelenke, so sind Repositionsversuche auf jede Fall zu unterlassen, da sie zu schweren Gefäß/Nerven Schäden führen können und da für eine erfolgreiche und dauerhafte Reposition auch nicht genügend kongruente Gelenkfläche zur Verfügung steht.

(F)

Le patient souffre d'une embryopathie due à la thalidomide; une **luxation congénitale** des épaules en fait partie. Il s'agit d'un status anatomique auquel le pat. est parfaitement adapté. Veuillez consulter les radiographies digitalisées incluses dans le dossier. Si les radiographies actuelles sont identiques à celles annexées, et, tant que le patient ne fait pas preuve d'une souffrance aiguë au niveau des épaules, il ne faut surtout pas procéder à une réduction de la luxation. L'anatomie des épaules que vous voyez est normale pour ce patient. La surface réduite de l'articulation ne permet pas un positionnement habituel; une tentative de repositionnement peut entraîner des complications neurovasculaires graves, sans autant bénéficier au patient.

(E)

El paciente presenta una embriopatía con **luxación congénital** de ambos hombros causado por talidomida. Se trata de una condición anatómica a la cual el paciente está perfectamente

adaptado. Para información adicional consulten las radiografías digitales anexados: Si no hay diferencia con los imágenes actuales, y al menos que el paciente se queja de molestias agudas en los hombros, una reducción de la luxación esta contraindicada. La situación anatómica que se presenta a ustedes esta normal para el paciente. La superficie reducida de la articulación de los hombros no permite un posicionamiento habitual. Una tentativa de reposicionamiento les hombro pueda causar graves lesiones neurovasculares, sin ningún beneficio para el paciente.

2.) Malformation of radial structures

(GB)

The patient has radial aplasia / dysplasia. For details of the patients bone structures of forearm and hand refer to the digital x-rays images included in the file.

(D)

Bei dem Patienten fehlt der radiale Strahl oder er ist fehlgebildet. Details zu den knöchernen Strukturen des Unterarmes und der Hand entnehmen Sie bitte den beiliegenden Röntgenaufnahmen.

(F)

Le patient est atteint d'une dysplasie / aplasie radiale. Pour plus de détails sur les structures osseuses de l'avant bras et de la main du patient, veuillez consulter les radiographies digitalisées inclus dans le dossier.

(E)

El paciente sufre de una displasia / aplasia radial. Para mas información sobre la estructura osea del antebrazo y de la mano del paciente, consulten las radiografías digitales anexadas .

3.) Missing radial arteria (Left arm)

(GB)

The patient suffers congenital aplasia of the radial artery on the left arm. Arterial puncture or insertion of arterial catheter can only be performed on the right arm.

(D)

Congenitale Aplasie der A. radialis links
Arterielle Punktionen im Bereich des Handgelenkes bitte nur rechts, da auf der linken Seite keine A. radialis angelegt ist.

(F)

Le patient souffre d'une anomalie vasculaire: Aplasie de l'artère radiale du bras gauche. Prise de sang (gasométrie, etc) artérielle dans la région du poignet peut seulement être effectuée sur la main droite.

(E)

El paciente sufre de una aplasia de la arteria radial del brazo izquierdo.. Manipulaciones arteriales (toma de sangre, cateterización arterial) a nivel de la muñeca deben hacerse solamente en la mano derecha.

3.) Missing radial artery (right arm)

(GB)

The patient suffers congenital aplasia of the radial artery on the right arm. Arterial puncture or insertion of arterial catheter can only be performed on the left arm.

(D)

Congenitale Aplasie der A. radialis rechts

Arterielle Punktionen im Bereich des Handgelenkes bitte nur links, da auf der rechten Seite keine A. radialis angelegt ist.

(F)

Le patient souffre d'une anomalie vasculaire: Aplasie de l'artère radiale du bras droite. Prise de sang (gasmétrie, etc) artérielle dans la région du poignet peut seulement être effectuée sur la main gauche.

(E)

El paciente sufre de una aplasia de la arteria radial del brazo derecho. Manipulaciones arteriales (toma de sangre, cateterización arterial) a nivel de la muñeca deben hacerse solamente en la mano izquierdo.

4.) Thoracic surgery in tetra-amelia.

(GB)

The patient has severe deformities of all four extremities. To maintain essential mobility and autonomy, the patient relies heavily on a stable and pain free thorax. If cardiovascular revascularization therapies are under discussion, we suggest that priority be given to the least invasive methods, such as thoracoscopic surgery or placement of stents. Open heart surgery would leave the patient completely helpless for weeks or months. Furthermore, due to the deformation of the legs, limited adequate vascular material for grafts has to be expected. A graft of the arteria mammaria may have to be considered as an alternative option.

(D)

Der Patient ist durch seine Vierfach Gliedmassenschädigung in seiner Beweglichkeit hochgradig eingeschränkt. Der Patient ist zur Wahrung seiner Restmobilität in höchstem Masse auf einen intakten und schmerzfreien Thorax angewiesen. Im Rahmen von cardialen Revaskularisationseingriffen ist zu erwägen, inwiefern nicht doch im Zweifelsfall einer Versorgung mit Stents bzw. einer thorakoskopischen Bypassversorgung vor einer offenen Chirurgie mit Sternotomie der Vorzug gegeben werden sollte. Weiterhin stehen an den geschädigten Beinen vermutlich nur eingeschränkt Blutgefäße zur Verfügung, die als Gefässinterponat bei einer Bypass Versorgung verwendet werden können. Im Zweifelsfall kann die Gewinnung eines A. mammaria Graftes sinnvoller sein.

(F)

Le patient est atteint de malformations des quatre membres (inferieurs et supérieurs). Pour sa mobilité et autonomie quotidienne, un thorax intact et indolore lui est indispensable. Si les thérapies de révascularisation cardiaques sont nécessaires, les méthodes peu invasives, telle que la chirurgie par thoracoscopie ou endoluminale doivent être considérées. Une intervention par sternotomie risque de laisser le patient démuné d'autonomie pendant des semaines, même des mois. De plus, il est important de rappeler, qu'une absence de matériels vaisseaux utilisables au niveau des jambes doit être prise en compte; cela est due aux malformations. Une greffe de l'arteria mammaria peut être considéré comme une alternative.

(E)

El paciente tiene malformaciones en los cuatro miembros (inferiores et superiores). Para su movilidad y autonomía cotidiana el paciente depende casi totalmente del tronco de su cuerpo; un tórax íntegro y sin dolor le es de una alta prioridad. En caso de verse necesario una terapia de revascularización cardíaca, hay que considerar en prioridad con métodos poco invasivos tales como la cirugía por toracoscópica o la angioplastia por cateterismo. Cirugía a corazón abierto con esternotomía dejará el paciente en situación de dependencia e inmovilidad durante semanas, posiblemente meses. Además, es posible que en los miembros inferiores malformados del paciente no haya ningún segmento vascular utilizable como injerto. En este caso la arteria mamaria puede considerarse como alternativa.