Rhabdoid tumours of the central nervous system (AT/RT) – Brief information

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1. General information on the disease

Rhabdoid tumours are rare, very aggressively growing *tumour*s, which mostly occur in infants and toddlers during the first two years of life. They are *embryonal* tumours, which means that they originate from extremely immature (*undifferentiated*) cells. Rhabdoid tumours can develop in any tissue of the body. Most frequently (with about 65 %), however, they affect *brain* and *spinal cord* (hence the *central nervous system*, CNS). Apart from that, rhabdoid tumours are found, above all, in the kidneys and the liver as well as in soft tissues (such as in the neck, thigh or chestwall area).

Rhabdoid tumours of the central nervous system (CNS) are also known as “atypical teratoid rhabdoid tumours" – briefly AT/RT. According to the current classification of the World Health Organisation (*WHO classification* CNS5), they are classified as grade 4 tumours (out of four possible grades), thus representing especially rapidly growing malignant *CNS tumour*s.

The following information is on rhabdoid tumours of the CNS only.

2. Incidence

Rhabdoid tumours of the central nervous system (AT/RT) are overall rare. In Germany, they account for about 0.6 % of all malignant diseases in children and adolescents under 18 years of age. This corresponds to an incidence rate of about 1 per million children. According to data of the European Registry for Rhabdoid Tumours (EU-RHAB), 15–22 patients per year have been newly diagnosed with AT/RT in Germany over the past years (*for more information on EU-RHAB, please see chapter “Therapy optimising trials and registries”*).

AT/RT appear in almost all age groups, however, infants and toddlers in their first two years of life are most frequently affected (with about 80 %). In children under one year of age, AT/RT are even the most common CNS tumours, according to data acquired by the German Childhood Cancer Registry (Mainz). From the age of six, the disease is rather rare. The average age at diagnosis is about 1.5 years. Boys are slightly less affected than girls (gender ratio: 0.9:1).

3. Site and spread

According to data of the EU-RHAB Registry (collected from 2005–2024), AT/RT develop, in about 50 % of cases, in the so-called *cerebellopontine angle* in the area of the *cerebellum* or the *brain stem* (that is, *infratentorial*ly), from where they invade into adjacent structures. About 34 % of

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the tumours are found above the *cerebellar tentorium* (*supratentorial*ly), for example within the hemispheres of the *cerebrum*. The remaining AT/RT distribute between the area of the *pineal gland*, the *diencephalon*, the *spinal cord*, or occur at multiple sites (multifocal). There are also tumours for which the exact primary site cannot be determined precisely due to their size. In older patients, supratentorial rhabdoid tumours occur more frequently than infratentorial rhabdoid tumours.

About 20–30 % of patients with AT/RT may present with metastases at diagnosis [see *metastasis*], predominantly within the central nervous system. Metastasis outside the *CNS*, on the other hand, is extremely rare and occurs almost exclusively in children with a predisposition for these tumours (a so-called rhabdoid tumour predisposition syndrome). In about 10–15% of patients with this *syndrome*, rhabdoid tumours also develop simultaneously in other parts of the body (multifocal, synchronous tumour involvement), which is not considered as metastasis.

4. Causes

The causes leading to the development of a rhabdoid tumour are not fully understood yet. A known fact, however, is that almost all (meaning over 90–95 % of) rhabdoid tumours – regardless of their localisation in the body – harbour a structural (*epigenetic*) alteration of a certain *gene* on *chromosome* 22. The affected gene is the *SMARCB1* gene (also known as *INI1*), which encodes the *protein* SMARCB1/INI1. This protein plays a major role in controlling cellular mechanisms such as cell growth and *differentiation*. The genetic defect (mutation) leads to loss of protein function and subsequently to predisposition to malignant transformation and tumour development.

Most of the time, the mutated *SMARCB1* gene is only found in the tumour cells following spontaneous malignant transformation of a somatic cell. Less frequently (in 25–30 % of patients), the *germline* (*germ cells*) and thus all cells in the body are affected (germline mutation). Both a spontaneous *genetic* change in the patient's germline during *embryonal development* and (very rarely) a defect inherited from a parent are considered as causes. In both cases the disease is hereditary, meaning that the mutated gene and thus the predisposition to develop a rhabdoid tumour can be passed on to the offspring. Experts communicate this as "**R**habdoid **T**umour **P**redisposition **S**yndrome" (RTPS). However, not all individuals harbouring a *SMARCB1* mutation will develop a rhabdoid tumour.

Aside from *SMARCB1* mutations, *SMARCA4* mutations (also known as *BRG1*) on chromosome 19 very rarely cause the tumour disease as well. This mutation is frequently inherited from a parent. Depending on which type of mutation has been found (*SMARCB1* oder *SMARCA4*), the syndrome is either called RTPS1 or RTPS2.

Good to know: In case of rhabdoid tumour predisposition syndrome (RTPS1 oder 2), thus a *germline mutation*, the patient's siblings are probably also at an increased risk of developing the disease. For both kinds of the syndrome are inherited in an autosomal dominant manner, which means that each child of a parent with RTPS has a 50 % risk of carrying the familial germline mutation as well. Therefore, genetic councelling for the patient's whole family is recommended when a rhabdoid tumour is suspected (*see chapter "Diagnosis"*).

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5. Symptoms

Due to the uncontrolled and aggressive growth pattern of rhabdoid tumours, *symptom*s typically develop and deteriorate within a few weeks. The presenting symptoms of these rhaboid tumours (AT/RT) (like other tumours of the *central nervous system*) primarily depend on the patient's age, the site and size of the tumour as well as its pattern of spread within the central nervous system (CNS). The following general (nonspecific) and local (specific) symptoms can occur:

5.1. General (nonspecific) symptoms

Unspecific general symptoms occur independently of the tumour's location. They may be similar to and therefore mimic other, non-CNS diseases. Infants and toddlers, as the most frequently affected age-group, typically present with shrill cries, irritability, fatigue, lethargy, loss of appetite, vomiting, developmental delay and/or failure to thrive. Wry neck (torticollis) as a result of cranial nerve impairment is also a common symptom. Older kids may complain about headaches and/or back pain as well as dizziness. General symptoms of a child or adolescent with a CNS tumour may also include increasing fatigue, inability to concentrate, school problems, mood swings, and character changes.

Major reason for these symptoms is the slowly but continuously increasing intracranial pressure (ICP). Elevated ICP may be caused by the growing, thus more and more space-occupying tumour within the bony skull, but also by the tumour blocking the regular flow of the *cerebrospinal fluid*, thereby forming *hydrocephalus*. In babies or small children with soft spots (open *fontanelle*s), elevated intracranial pressure and hydrocephalus typically present with a bulging fontanelle or a larger than expected head circumference (*macrocephalus*), respectively.

5.2. Local (specific) symptoms

Local symptoms may indicate the tumour location and, thus, which functional regions of the CNS may be affected. Therefore, a rhabdoid tumour in the *cerebellum* can cause impaired movement, visual deficits, dizziness and gait disturbances, whereas such a tumour in the hemispheres (*cerebrum*) can be associated with motor and sensory deficits (one-side paralyses, hemiplegia), *seizures*, speech disorders and behavioural problems, to name a few. A tumour in in the *spinal cord* area may cause serious back pain as well as palsies of various types (such as bladder and bowel incontinence).

Good to know: Not all patients presenting with one or more of the symptoms mentioned above do have a rhabdoid tumour or another type of brain tumour. Many of these symptoms may also occur with other, harmless diseases that are not associated with a brain tumour at all. However, if certain symptoms persist or get worse (for example repetitive headaches or rapid increase of head circumference in a young child), a doctor should be seen to find the underlying reason. In case it is a rhabdoid tumour or some other brain tumour, treatment should be started as soon as possible.

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6. Diagnosis

If the paediatrician thinks that the young patient's history (*anamnesis*) and *physical examination* are suspicious of a tumour of the *central nervous system* (CNS), the child should immediately be referred to a hospital with a childhood cancer program (paediatric oncology unit), where further diagnostics can be initiated and performed by childhood cancer professionals. Very close collaboration between various specialists (such as paediatric oncologists, paediatric neurosurgeons, paediatric radiologists, to name a few) is required, both to find out, whether the patient really suffers from a malignant *CNS tumour* and, if so, to determine the tumour type and the extension of the disease. Knowing these details is absolutely essential for optimal treatment and *prognosis*.

6.1. Imaging tests to confirm tumour existence

The initial diagnostic procedures for a young patient presenting with a suspected CNS tumour at a childhood cancer centre include another assessment of the patient's history, a thorough physical/ *neurological* exam and *imaging* diagnostics, such as *ultrasound* (sonography) and *magnetic resonance imaging* (MRI). Rapid clarification may require *computed tomography* (emergency CT). These tests help to find out exactly whether the patient has a tumour of the central nervous system. Also, localisation and extent of the tumor as well as its demarcation regarding adjacent tissue can be diagnosed by these imaging techniques very well.

6.2. Tissue extraction (biopsy) to secure diagnosis

Final diagnosis of a CNS tumour requieres the removal of tumour tissue (*biopsy*). This usually requires surgery. The obtained tissue will be analysed under the microscope (histologically) as well as with *immunohistochemical*, *cytogenetic* and *molecular genetic* methods. In particular, proof of *SMARCB1* or *SMARCA4 mutation* facilitates establishing the diagnosis of a rhabdoid tumour (*see chapter "Causes"*). In the context of immunohistochemical analysis, loss of the SMARCB1 or SMARCA4 protein as a result of the mutation can be determined by specific staining of the tumour cells. Cytogenetic and molecular genetic testing provides direct proof of the genetic defect.

Once a *SMARCB1* or *SMARCA4* mutation has been confirmed for the tumour tissue, blood cells (or any other tumour-free tissue) will be tested for the mutation as well in order to rule out a *germline mutation* and rhabdoid tumour predisposition syndrome (RTPS), since in case of a germline mutation, the blood cells (or other tissue cells) do contain the mutation as well. The germline mutation is particularly suspected in children under two years of age, in patients with tumours at multiple sites (synchrone tumours) or in case of a family history of tumour diseases, respectively.

In case a family history of rhabdoid tumour predisposition syndrome (RTPS1 or RTPS2) is known prior to biopsy, testing only the patient's blood for mutation can be sufficient to confirm diagnosis, since a malignant tumour revealed by diagnostic imaging and with *SMARCB1* or *SMARCA4* mutation in the patient's blood cells is most likely an AT/RT. However, blood tests cannot replace biopsy and may, therefore, be an option but in very rare situations (for example, if the child is to ill to be operated).

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Aside from the examination of tumour tissue for a *SMARCB1* or *SMARCA4* mutation, the determination of the *DNA methylation* profile is becoming increasingly important. With the help of this molecular genetic method, AT/RT can be divided into three different *molecular* (*epigenetic*) subtypes (ATRT-MYC, ATRT-SHH, ATRT-TYR), some of which also behave differently regarding their growth pattern (*see chapter "Therapy planning"*).

Important to know: diagnosis of a germline mutation not only affects a patient's *prognosis*, but maybe also other family members, especially regarding the need for subsequent screening tests (*see recommendations in case of a rhabdoid tumour predisposition syndrome below*).

6.3. Tests to assess spread of disease (tumour staging)

Once the diagnosis of a rhabdoid tumour has been confirmed, additional tests are required to assess the extent of the disease within body (stage of disease), Since rhabdoid tumours are known to metastasize frequently and also may be found at multiple sites already at diagnosis, for example in the central nervous system (CNS) and simultaneously in the kidneys (synchronous rhabdoid tumour), the initial finding of a single tumour is always followed by diagnostic imaging of the whole body.

Usually, a *magnetic resonance imaging* (MRI) of the complete CNS (brain and spine) is performed to rule out *metastasis* within the CNS, furthermore, at least once, a total body scan to assess macroscopic distant metastases and/or synchronous rhabdoid tumours. Apart from that, mostly after surgery, the *cerebrospinal fluid* (CSF) is microscopically checked for tumour cells in the spinal cord (which are not visible by MRI scan). Cerebrospinal fluid is mostly obtained from the spine in the lower back (*lumbar puncture*), since the risk of the puncture needle damaging the spinal cord is lowest at the lower back level.

6.4. Tests before treatment begins

In preparation for the intensive treatment of the brain tumour, further investigations are performed, such as *electrocardiography* (ECG) and/or*echocardiography* to check cardio function or *electroencephalography* (EEG) to test brain function. Furthermore, blood tests are needed to assess the patient's general health condition and to check whether the function of certain organs (such as liver and kidneys) is affected by the disease and whether there are any metabolic disorders to be considered prior or during therapy. Any changes occurring during the course of treatment can be assessed and managed better based on the results of those initial tests, which thus help to keep the risk of certain treatment-related side effects as low as possible.

If the child's general condition is stable, neuropsychological examinations (for example by means of questionnaires) can also be carried out at the time of diagnosis [see *neuropsychology*]. These serve to record the child's level of development and *cognitive* abilities as well as any abnormalities as a basis for future surveys. Examinations on quality of life may also be included.

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Good to know: Not every patient needs the complete check-up. On the other hand, tests might be added that haven't been mentioned here, depending on the individual situation of the patient. Your caregivers will inform you and your child, which diagnostic procedures are individually required in your case and why.

6.5. Recommendations if a rhabdoid tumour predisposition syndrome is suspected

If a patient is diagnosed with a *SMARCB1 germline mutation* – and, subsequently, is at an increased risk for developing a rhabdoid tumour (rhabdoid tumour predisposition syndrom, RTPS) –, one possible reason may be that the disease has been inherited by one parent. In most of the cases, however, it is more likely that the genetic defect is a result of a new *SMARCB1 mutation*.

If inheritance is the case, both the patient's parents and siblings are at a higher risk to develop a rhabdoid tumour as well (*see chapter „Causes"*). In order to rule out this risk or to diagnose it as early as possible, certain blood tests will be recommended to both parents first. If those come back positive for a germline mutation in one of the parents, the patient's siblings should also be tested. These tests are usually done in human genetics laboratories. Also, genetic councelling is recommended.

If a rhabdoid tumour predesposition syndrome is known in the family and a germline mutation is found in a so far healthy family member, it is recommended that the affected child(ren) is (are) closely monitored from birth by regular physical/*neurological* examinations, *magnetic resonance imaging* and *ultrasound* (of the head, abdomen and chest).

Psychosocial Care

A child's cancer is a stressful situation for the whole family. The psychosocial team of the clinic or later the aftercare facility provides advice and support to patients and their relatives from diagnosis to completion of treatment as well as during aftercare. Don't hesitate to take advantage of this offer. It is an integral part of the treatment concept of all paediatric oncology centres in many countries. Here you will find comprehensive information on this.

7. Treatment planning

After the diagnosis has been confirmed, therapy is planned. In order to design a highly individual, risk-adapted treatment regimen for the patient, certain individual factors influencing the patient's *prognosis* (called risk factors or *prognostic factors*) are being considered during treatment planning (risk-adapted treatment strategy).

One important prognostic factor is the patient's age at diagnosis: it determines treatment intensity and, therefore, has an impact on the patient's chances of survival. For example, radiotherapy – a very efficient treatment method for rhabdoid tumours – is only a limited option for children under the age of three and no option at all for children under the age of 12 months. Also, the tolerance of this treatment is significantly reduced in very young children when compared to other methods (such as surgery, chemotherapy).

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Another important prognostic factor is the *molecular* subtype of AT/RT, since this may also impact the course of the disease (s*ee chapter "Diagnosis / Tissue extraction"*). For example, patients with the ATRT-TYR subtype usually have a better chance of cure than patients with one of the other two subtypes (ATRT-MYC, ATRT-SHH). In summary, knowledge of the subtype allows drawing conclusions on prognosis.

Localization, size and spread of the tumour as well as its type (hereditary or non-hereditary) are further factors affecting prognosis. Hereditary disease (that is, a rhabdoid tumour predisposition syndrome, RTPS), is known as an unfavourable prognostic factor and so is *metastasis* at the timepoint of tumour diagnosis. Both scenarios are associated with a lower probability of eliminating all tumour manifestations (which is known to promote favourable prognosis). Response of the disease to chemotherapy is also an important prognostic factor.

All these factors are included in treatment planning in order to achieve the best outcome possible for each patient.

8. Treatment

Treatment of children and adolescents with rhabdoid tumour should take place in a children's hospital with a paediatric oncology program. Only such a childhood cancer centre provides highly experienced and qualified staff (doctors, nurses and many more), since they are specialised and focussed on the diagnostics and treatment of children and teenagers with cancer according to the most advanced treatment concepts. The doctors in these centres collaborate closely with each other. Together, they treat their patients according to treatment plans (protocols) that are continuously optimised.

The goal of the treatment is to achieve high cure rates while avoiding side effects as much as possible. Considering the usually very young age of patients with a rhabdoid tumour, this is a huge challenge. Infants and toddlers are highly vulnerable; they suffer severely of acute side effects and long-term sequelae of the aggressive therapy and are therefore hard to treat. Hence, the most important step prior to or during therapy is to decide whether treatment should be initiated or continued, respectively, and if so, whether the goal is cure (curative approach) or managing symptoms (*palliative therapy*).

From 2007 until the opening of the international therapy trial SIOPE ATRT01 in 2021, patients with rhabdoid tumour received treatment according to a standardized therapy protocol under the roof of the EU-RHAB Registry (so-called consensus treatment strategy, *see chapter "Therapy optimising trials and registries"*). The following information on therapy is based on this consensus treatment strategy and the current guidelines of the Society of Paediatric Oncology and Haematology (*GPOH*) for AT/RT.

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8.1. Treatment methods

Treatment options for patients with rhabdoid tumour include **surgery**, **chemotherapy** and **radiotherapy**. For some patients, **high-dose chemotherapy** followed by **autologous stem cell transplantion** may be an option, too. The individual treatment choice is based on the patient's age and general health status as well as on the tumour type and its extent at diagnosis and after surgery.

Neurosurgery and *radiotherapy* are the most promising therapy methods and of highest impact in the treatment of patients with rhabdoid tumour. Yet, *surgery* or radiation cannot be performed in every child. Radiotherapy, for example, is only possible from a certain age. Chemotherapy (and, for some patients, *high-dose chemotherapy*) can help improve prognosis and – particularly in very young children – delay or even completely avoid radiotherapy.

8.1.1. Surgery

First step when treating a patient with rhabdoid tumour is maximal neurosurgical tumour removal [see *neurosurgery*], if possible, since the extent of tumour resection seem to have a major impact on the subsequent course of the disease. The more radically the tumour can be resected, the higher are the chances of long-term survival. If the tumour is localized and has not spread, second look surgery to achieve gross total removal is an option. Unfortunately though, complete resection is impossible in most cases (in about 70 % of patients) without also removing healthy tissue and without jeopardizing the patient's quality of life. This is related to the frequently unfavourable tumour location, the patient's young age as well as the frequent presence of metastases [see *metastasis*] at the timepoint of tumour diagnosis.

Aside from tumour removal, neurosurgery also includes the implantation of a *catheter* into one of the brain's ventricles (*cerebral ventricles*) for subsequent chemotherapy. The catheter is connected to a small, dome-shaped, soft plastic device (*Ommaya reservoir* or *Rickham reservoir*) placed under the patient's scalp, through which chemotherapy can directly be delivered into the *cerebrospinal fluid* (*see next chapter*).

8.1.2. Chemotherapy

Surgery is followed by intensive *chemotherapy* in order to improve the patient's chances of cure. Chemotherapy uses drugs (so-called cytostatic agents or cytostatics) that can kill fast-dividing cells, such as cancer cells, or inhibit their growth, respectively. In order to optimize treatment efficacy, combinations of different *cytostatics* are given in different treatment blocks.

Standard therapy according to the current guideline consists of two phases, an induction and a consolidation phase, and may include up to 12 treatment blocks. Induction therapy aims at a maximum possible control of the tumour and/or the tumour cell count, while the aim of consolidation therapy is to maintain (consolidate) the results gained by induction. The therapy includes the agents doxorubicine (DOX) and combinations of ifosfamide, carboplatin and etoposide (briefly: ICE) or vincristine, cyclophosphamide and actinomycin D (briefly: VCA), respectively, which are given *intravenous*ly in alternation.

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By the intravenous route, they get distributed in the blood system and can, thus, eliminate tumour cells throughout the whole body (*systemic* chemotherapy). In addition to this approach, some chemotherapy (methotrexate, MTX) is given directly into the *cerebrospinal fluid*, which surrounds both brain and spinal cord. This intraventricular or *intrathecal chemotherapy* is necessary, because most chemotherapeutic agents cannot pass the barrier between blood and brain (*blood-brain barrier*).

8.1.3. Radiotherapy

Depending on the patient's age at the timepoint of treatment, *radiotherapy* may be recommended during or after chemotherapy. Radiotherapy is carried out using energy-rich, *electromagnetic* radiation, given through the skin to the tumour region. Radiation causes *DNA* damage in tumour cells, thereby leading to cell death.

Aside from complete surgical tumour removal, radiotherapy is one of the most important and successful measures for treatment of a rhabdoid tumour. Studies have shown that patients benefit from early radiation. However, its application is limited due to treatment-induced late effects. This particularly affects infants and toddlers, whose brains are still developing: radiation early in life results in serious impairment of normal *cognitive* development. Therefore, chemotherapy and, if applicable, even *high-dose chemotherapy* are used to delay radiotherapy as long as possible.

If radiotherapy is an option, timing, volume and type (*photon*s versus *proton*s) are determined based on the patient's age, the vulnerability of the tissue and prognostic factors.

According to the current guidelines for AT/RT, radiotherapy is considered for children 12–18 months of age with localized disease. These patients will receive a total dose of 54 *gray* (Gy) to the tumour region. For children whose tumour has spread within the central nervous system via the *cerebrospinal fluid* (leptomeningeally metastasized tumours), radiation of the brain and spinal cord (craniospinal radiotherapy) are considered once they are three years old. Fore these patients, a dose of 35.2 Gy to the whole central nervous system plus an additional dose (boost) to the *primary tumour* site (up to a total dose of 55 Gy) are recommended.

Modern radiation techniques, such as *intensity-modulated radiotherapy* (IMRT), help minimise the damage of healthy tissue. For some patients, radiotherapy with protons instead of conventional radiotherapy (with photons) may be an option, for example for very young children or in case *proton therapy* is expected to have a clear advantage compared to conventional radiotherapy. This type of radiotherapy allows to reduce the effects of radiation in healthy tissue even better and is, therefore, gaining an increasing importance in the treatment of children and teenagers with solid tumours.

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Note on the SIOPE ATRT01 trial: This study examines particularly, whether radiation-induced long-term sequelae in young children can be avoided by the use of high-dose chemotherapy only (no radiotherapy) without jeopardizing the patient's chances of cure. Hence, the feasibility of high-dose chemotherapy only versus the current regimen of combined chemo- and radiotherapy (12 courses of chemotherapy plus radiotherapy) will be analysed. The study will include children aged 12–35 months who have no metastases and no rhabdoid tumour predisposition syndrome (RTPS).

8.1.4. High-dose chemotherapy and autologous stem cell transplantation

For some patients, *high-dose chemotherapy* as a consolidation treatment followed by *autologous stem cell transplantation* may be an option instead of the conventional chemotherapy described above. In this case, the patient receives treatment with carboplatin and thiotepa (CARBO/TT) after six cycles of standard chemotherapy (induction therapy, *see chapter "chemotherapy"*). The chemotherapy doses of this regimen are considered high enough to also eliminate otherwise treatment-resistant tumour cells in the body.

Since the high doses of *cytostatics* given according to this treatment strategy also lead to the destruction of the blood-forming cells in the *bone marrow*, the patient will receive blood-forming stem cells in a second step. These *blood stem cells* are obtained from the patient's blood or bone marrow prior to high-dose chemotherapy and are given back right after this treatment (so-called autologous stem cell transplantation, SCT).

Precondition for carrying out this treatment is, however, that most of the tumour load has been destroyed by the preceding standard regimen, thereby having achieved a so-called *remission*. Also, the patient's age and general clinical condition are of relevance, for the treatment will be stressful and associated with risks.

It is still unclear so far whether the high-dose regimen provides a prognostic benefit over the standard chemotherapy (in children under 12 months of age) or the conventional chemo-/ radiotherapy (in children older than 12 months). This question is currently being addressed by the European Umbrella trial SIOPE ATRT01, which has been open since 01/06/2021.

Note on the SIOPE ATRT01 trial: The study will examine whether high-dose chemotherapy (without radiation) for consolidation achieves the same results as combined radiotherapy/ chemotherapy in children aged 12–35 months – following induction chemotherapy. For children under 12 months of age or children who are not eligible for radiotherapy, the study will examine whether high-dose chemotherapy leads to better survival rates than continuation of conventional chemotherapy in the consolidation.

For further, general information on stem cell transplantation, please see *here*.

8.1.5. New treatment approaches

Despite the currently available intensive treatment modalities, cure rates for children with AT/RT are unsatisfying. This particularly applies to high-risk patients (young age at diagnosis, unfavourable

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molecular subtype, *germline mutation* and/or metastasised tumour). In addition, the intensive treatment does not only cause acute side effects, but also long-term sequelae (such as hormonal deficits, which are associated with certain developmental delays, or impairments in hearing or vision). This may seriously impair the patients' quality of life.

Scientists keep studying these tumours intensely to find new agents and treatment modalities. The current research focuses on the molecular mechanisms leading to the developments and growth of rhabdoid tumours. The analysis of cellular signalling pathways that are altered in rhabdoid tumours have helped identifying different agents, which may be of benefit in the treatment of rhabdoid tumours. Promising new treatment strategies will be examined in the framework of clinical studies.

For patients with a relapse of an AT/RT, various approaches apply to individual treatment attempts. These include *epigenetic*ally active agents like decitabine on the one hand, but also the drug ribociclib and so-called *checkpoint inhibitor*. Another therapy option is a *metronomic therapy*s, for example within the framework of the MEMMAT protocol.

Unfortunately, there are currently only few phase I/II studies open for children. The NivEnt study, for example, only includes children with a certain molecular AT/RT subtype (ATRT-MYC). The researchers at the EU-RHAB Registry are working hard to find new drugs and get them approved for clinical use. This should always be carried out as clinical studies (*see also next chapters*).

9. Therapy optimising trials and registries

In the large paediatric treatment centres, children and teenagers with rhabdoid tumour receive therapy according to guidelines and/or standardized treatment plans (protocols). These protocols are designed by experts with the goal to improve the patient's *prognosis* and are usually applied within *therapy optimising trial*s or registries. Therapy optimising trials are standardised and controlled clinical trials that aim at steadily developing and improving treatment concepts for sick patients based on the current scientific knowledge.

Patients who cannot participate in any study, for example because none is available or open for them at that time, or since they do not meet the required inclusion criteria, respectively, are often included in a so-called **registry**. Such a registry primarily serves to acquire all clinical, *molecular genetic* and treatment-associated patient data in ordert o gain a better understanding oft he tumour biology. Furthermore, the registry center supports the doctors at site with (non-commital) treatment recommendations based on the most recent data on best treatment options, in order to provide the patient with optimal therapy even without the framework of a clinical study.

Currently available for patients with AT/RT are the trial SIOPE ATRT 01 and the EU-RHAB registry. Further trials are being developed. *For more information on trials and registry please see below.*

9.1. European Rhabdoid Registry (EU-RHAB Registry)

Since rhabdoid tumours are very rare, experts of the Society of Paediatric Oncology and Haematology (*GPOH*) initiated during a consensus conference in Italy in 2007 that all patients with

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rhabdoid tumours in a part of Europe should be registered with the European Rhabdoid Registry (EU-RHAB Registry) and treated according to a standardized strategy (consensus strategy). The treatment regimen was designed by an international competence network and is outlined in the chapter “Treatment” as well as in the current guideline for AT/RT. It has been validated as standard therapy, unless the patient is not treated within the experimental arm of a trial, and requires adjustment to the individual patient.

Numerous countries worldwide are currently registering their patients with this EU-RHAB Registry. Treatment centres in Germany, where children and adolescents with cancer are being treated, are legally obliged to register their patients with the appropriate studies or registries. The European Registry headquarters are located at the University Hospital in Augsburg, Germany, and the principal investigator is Prof. Dr. Dr. med. Michael C. Frühwald. **Please note:** Patient recruitment has been stopped on a temporary basis since 24/04/2023! A new registry (EU-RHAB 2.0) will be opened shortly.

9.2. Therapy trial SIOPE ATRT01

In July 2021, an international, multicentric trial – developed in the framework of the EU-RHAB registry – has been opened for children with AT/RT: the umbrella study SIOPE ATRT01. In this context, the term "umbrella" means that the study consists of several parts and assigns the affected patients to a therapy arm depending on their individual risk profile.

The umbrella study enrolls all European patients aged 0–17 years. The integrated randomised *therapy optimising trial* is intended for patients aged 12–35 months [see *randomisation*]. Numerous treatment centres in Germany and in almost all other European countries (including France, Switzerland, England, Spain and Portugal) are participating in the study. The study centre is located at the University Hospital in Augsburg, Germany (principal investigator: Prof. Dr. Dr. med. Michael C. Frühwald).

9.3. New trials

Within the framework of EU-RHAB, new studies are also being developed based on previously acquired data. A Phase I/II study is currently under development to test a new substance that is presumably effective in rhabdoid tumours. In addition, a new treatment protocol is being developed for the treatment of patients with disease relapse.

10. Prognosis

The chances of cure (*prognosis*) for children with a newly diagnosed rhabdoid tumour of the central nervous system have significantly improved thanks to standardised, multimodal treatment concepts within the framework of EU-RHAB. Patients treated in the most favourable risk (therapy) group meanwhile present with 5-year-survival rates of more than 71.5 %. For patients with unfavourable prognostic factors, however, prognosis is still not favourable, despite the intensive therapy. According to data of the EU-Rhab Registry, the average overall 5-year-survival rate (for all risk groups taken together) is about 35–40 %.

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Important prognostic factors are the patient's age at diagnosis, tumour type (hereditary or non-hereditary, *molecular* subtype) as well as tumour size, site and extent and, thus, possibility of complete tumour removal (*see chapter "Treatment planning"*). Survival rates are thus different in individual patients:

- Patients with localized, non-metastasised, surgically removable and non-hereditary rhabdoid tumour, who are older than three years of age at diagnosis, usually have favourable probabilities of cure, given that the tumour can be completely removed and early radiotherapy is possible.
- Children between their first and third birthday with the same co-factors have a comparably higher risk of relapse and thus less favourable outcomes.
- Prognosis is particularly unfavourable for infants and toddlers under one year of age, especially if they present with a certain molecular AT/RT subtype.
- The same applies to all other patients with unfavourable prognostic factors, that is, a high-risk rhabdoid tumour. These include patients with *germline mutation*, and thus a predisposition for the development of rhabdoid tumour, as well as patients with surgically unremovable *primary tumour* or metastasised disease, respectively.

However, patients who do benefit from treatment (surgery, chemotherapy, in some cases high-dose chemotherapy and radiotherapy) despite of unfavourable prognostic factors, so that long-term survival may be possible. New molecular treatment approaches are currently being analysed in the frame-work of therapy optimising trials. The goal is to optimize cure rates also for high-risk patients.

Note: The survival rates mentioned in the text above are statistical values. Therefore, they only provide information on the total cohort of patients with this kind of brain tumours. They do not predict individual outcomes.

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Glossary

anamnesis	medical interview, a patient's history, development of signs of illness; the type, onset and course of the (current) symptoms as well as any risk factors (e.g. hereditary diseases) are evaluated during a medical interview.
autologous stem cell transplantation	(re)transfer of blood stem cells, e.g. after a chemotherapy or radiotherapy; the patient receives his own cells that were previously taken from their own bone marrow or blood. Autologous stem cell transplantation may be an option, for example, for certain patients with lymphoma, neuroblastoma, soft tissue sarcoma, or a brain tumour.
biopsy	removal of a tissue sample for subsequent (mainly microscopic) examination; this can be done, for example, by puncture with a hollow needle, with the use of special instruments (such as forceps, punching instruments, probes) or surgically with a scalpel.
blood stem cells	precursor cells of all blood cells, which give rise to red blood cells (erythrocytes), white blood cells (leukocytes), platelets (thrombocytes) and some other cells. This process is called blood formation. The various blood cells are formed in the bone marrow before they enter the blood stream.
blood-brain barrier	barrier between the blood and the central nervous system (CNS) that is permeable only to certain endogenous and foreign substances, thereby enabling active control over the exchange of substances with the CNS;
bone marrow	site of blood formation; spongy tissue with a strong blood supply that fills the cavities inside many bones (e.g., vertebrae, pelvic and thigh bones, ribs, sternum, shoulder blade, and collarbone); in the bone marrow, all forms of blood cells develop from blood progenitor cells (blood stem cells).
brain	the part of the central nervous system (CNS) located in the head; the brain is protected by the skull and the meninges and consists mainly of nerve tissue.
catheter	tubular, rigid or flexible instrument for insertion into hollow organs, vessels or specific body cavities (e.g. bladder), e.g. for examination, drainage, sample collection, monitoring of vital parameters and/or administration of medication

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central nervous system	comprises the brain and spinal cord and is separated from the so-called peripheral nervous system; as a central organ of integration, coordination and regulation, it serves to process external sensory impressions as well as stimuli produced by the organism itself.
cerebellar tentorium	connective tissue structure that separates the posterior parts of the cerebrum from the cerebellum like a kind of parting sail, leaving only one passage route for the brainstem; it is formed by the hard meninges and covers the posterior cranial fossa like a roof.
cerebellopontine angle	niche in the posterior region of the brain and part of the cerebellum; this is where the roots (nuclei) of ten of the twelve cranial nerves are located in a very small space.
cerebellum	part of the brain that is located in the posterior fossa of the skull, between the cerebrum and the brainstem; it is mostly responsible for the coordination of all body movements and also for maintaining balance.
cerebral ventricles	cerebral ventricles filled with cerebrospinal fluid; the four cerebral ventricles represent the continuation of the spinal canal merging into these four chambers in the brain.
cerebrospinal fluid	fluid produced by cells of the cerebral ventricles; it floats around the brain and spinal cord to protect them from injury and provide them with nutrients.
cerebrum	largest and most highly developed section of the brain; it consists of two hemispheres connected by a thick bundle of nerves (corpus callosum). Each hemisphere of the brain is specialized on specific tasks. The outermost layer of the cerebrum, the cerebral cortex, houses the ability to learn, speak and think, as well as consciousness and memory, amongst other things. This is also where the processing centres for information from the sensory organs (e.g. eyes, ears) are located.
checkpoint inhibitor	drug (monoclonal antibody) that activates the bodys own immune response against tumour cells; is used specifically as immunotherapy in cancer treatment. The effect of checkpoint inhibitors is based on the blocking of certain switching points of the immune system, so called immune checkpoints. These are surface proteins on immune cells (T lymphocytes) that ensure that an immune reaction is terminated on time. This checkpoint control usually serves to ensure that the immune system does not react too strongly or is directed against the bodys own tissue

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	(autoimmune reaction). Cancer cells can also activate these checkpoints and thus slow down the bodys immune response. By blocking these switching points, checkpoint inhibitors reactivate the immune cells, thereby initiating an immune response against the tumour cells.
chemotherapy	here: use of drugs (chemotherapeutic agents, cytostatics) for the specific inhibition of tumor cells in the organism
chromosome	carriers of the genetic material, i.e. the genetic information of a cell; chromosomes consist mainly of DNA and proteins and are components of the cell nucleus. The shape and number of chromosomes are species-specific. Humans have 46 chromosomes (23 pairs of chromosomes) per cell in the body.
chromosome	
CNS	abbreviation for central nervous system
CNS tumour	tumour of the central nervous system; a primary CNS tumour is a solid tumour that originates from brain or spinal cord tissue. Secondary CNS tumours are metastases of tumours located in other organs or tissues.
cognitive	This term refers to the information-processing processes that take place in the brain. Cognitive processes include thinking, memory, decision-making, and cognition. Cognitive functions involve, for example, perception, attention, concentration, memory, action planning, judgment, problem solving and communication.
computed tomography	imaging, X-ray diagnostic procedure; it produces an image by computer-controlled evaluation of a large number of X-rays from different directions. This makes it possible to produce sliced images of body parts (tomograms, transverse or longitudinal sections of the human body)
cytogenetic	the number and structure of the chromosomes contained in the nucleus
cytostatics	drugs that inhibit cell growth; cytostatics can affect the metabolism of different types of cells, thereby destroying them and/or preventing them from multiplying. Cells that divide frequently are particularly affected.
diencephalon	vital part of the brain with function for numerous life processes; it connects to the brainstem towards the cerebrum and consists of functionally different sections. The "thalamus", for example, decides which sensory impressions should penetrate into

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	consciousness and are to be sent to the appropriate processing centers. The "hypothalamus" serves as a mediator between the hormonal and nervous systems and controls, among other things, important metabolic processes (e.g. heat and water balance, carbohydrate, fat, protein metabolism, blood pressure). Together with the pituitary gland, it regulates the activity of subordinate glands. Other parts of the diencephalon are responsible for muscle activities and for controlling the day-night rhythm.
differentiation	here: development of immature cells / immature tissue into mature tissue with specialised tasks; differentiation is based on a hereditary blueprint.
DNA	abbreviation for deoxyribonucleic acid; it carries the genetic information and is found in all living beings. DNA contains the genes that provide the information for the production of ribonucleic acids (RNA) or proteins. It is a large molecule consisting of two nucleic acid chains twisted into a double helix. The individual chains consist of a sequence of four different building blocks (bases), the order (sequence) of which determines the genetic code.
DNA methylation	regulatory process that controls the activity of genes (epigenetic regulation); in this process, methyl groups are coupled by enzymes with certain building blocks (nucleotides, nucleobases) of DNA, thereby chemically modifying them. Since the basic structure of the nucleotides is preserved, this is not causing mutation, but an epigenetic change (modification). Defective DNA methylation is usually passed on to daughter cells and is therefore often the cause of diseases. The epigenetic activation or suppression of genes by means of methylation can contribute to the development of cancer.
echocardiography	ultrasound examination of the heart to check its performance (cardiac function); the position or structure of the heart valves and walls, the wall thickness of the heart muscle, the size of the heart and the ejected blood volume (pumping function of the heart) are examined and assessed, among other things.
electrocardiography	method of measuring the electrical activity of the heart
electroencephalography	method of recording the electrical activity of the brain; the electroencephalogram (also abbreviated EEG) is the graphical representation of this electrical brain activity. Its evaluation can provide evidence of brain dysfunctions.

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electromagnetic	electromagnetic rays (also known as electromagnetic waves) consist of coupled electric and magnetic fields; examples of electromagnetic radiation are X-rays and gamma rays as well as radio waves, thermal radiation and light.
embryonal	here: in an early stage of development, immature;
epigenetic	epigenetics is a branch of biology that deals with molecular mechanisms that lead to stronger or weaker expression of genes without altering the information stored on the gene. Instead, certain biocatalysts (enzymes) mark certain sections on the genetic material (DNA). In contrast to genetic processes, epigenetic processes are reversible and do not influence the sequence of the DNA, but the way the sequence is being read by taking place on top of it, i.e. at a higher level ("epi-" - from Greek: "over"). Epigenetic processes are nevertheless heritable, meaning they are passed on during cell division. Through epigenetics, cells control, for example, which proteins they produce, in what quantities and when.
fontanelle	soft spot on an infant’s head, due to the bony plates not having connected yet; the final closure usually occurs before the age of two.
gene	unit of genetic information in the genome of living organisms; a gene contains the genetic information – the blueprint – for a specific gene product (protein or RNA). In most organisms, the entirety of all genes, the genome, is present as a deoxyribonucleic acid chain (DNA), which forms the chromosomes in the cell nucleus. The information of a gene is mediated by a certain sequence of the nucleic acid building blocks adenine, guanine, cytosine and thymine.
genetic	concerning the (level of) inheritance or genes; inherited
germ cells	mature cells capable of sexual fertilization (eggs in women, sperm cells in men)
germline	term for those cells that are in the service of the direct transmission of genetic material, i.e. from which germ cells (egg cells and sperms) arise in the course of individual development; the germline begins with the fertilized cell (zygote) and continues through the formation of primordial germ cells to the formation of the sex glands (gonads), which are responsible for reproduction and ultimately the germ cells.

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germline mutation	mutation that occurs in the female or male germ cells (eggs or sperm) and can thus be inherited by the offspring; in the case of a germline mutation, all body cells of the offspring are usually affected by the change. In contrast, "somatic" mutations arise in somatic cells outside the germline and are not inherited.
GPOH	Society for Paediatric Oncology and Hematology (GPOH), the German professional society for childhood and adolescent cancers and blood diseases; in the GPOH, doctors, scientists, nurses and psychologists, among others, work together on the research, diagnosis, treatment and aftercare of malignant diseases and blood diseases in children and adolescents. in the GPOH, doctors, scientists, nurses and psychologists, among others, work together on the research, diagnosis, treatment and aftercare of malignant diseases and blood diseases in children and adolescents.
gray	unit of measurement for the dose of energy caused by ionising radiation (e.g. in the context of radiotherapy) and absorbed by a given mass (kilogram of body weight)
high-dose chemotherapy	the use of a particularly high dose of cell growth-inhibiting drugs (cytostatics); in the case of cancer, it aims to destroy all malignant cells. Since the haematopoietic system in the bone marrow is also destroyed, the patients own or foreign blood stem cells must then be transferred (autologous or allogeneic stem cell transplantation).
hydrocephalus	medical term for abnormal buildup of cerebrospinal fluid in the cavities (ventricles) in the brain; it is caused by a dilation of the brain’s ventricles due to various causes.
imaging	diagnostic procedures generating images of the inside of the body, such as ultrasound and X-ray examination, computed tomography, magnetic resonance imaging, and scintigraphy
immunohistochemical	in an immunohistochemical or immunohistological examination, proteins or other cell or tissue structures are visualized with the help of labeled antibodies (e.g. bound to dyes).
infratentorial	located below the cerebellar tentorium, i.e. in the posterior cranial fossa
intensity-modulated radiotherapy	modern radiation technology, which provides maximum protection for the surrounding healthy tissue from radiation exposure by means of a highly precise distribution of the radiation dose at the tumour site; the intensity of the radiation dose can be precisely

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	adjusted to the irradiation field only; this may also allow the use of a higher radiation dose.
intrathecal chemotherapy	administration of cell growth-inhibiting drugs (cytostatics) into the cerebrospinal fluid (CSF) canal, which contains the CSF
intravenous	means located within a vein or given into a vein; here: e.g. administration of a medication or fluid/suspension into the vein by an injection, infusion or transfusion.
lumbar puncture	puncture of the spinal canal in the lumbar spine, e.g. to remove cerebrospinal fluid (CSF) or for the purpose of administering medication (so-called intrathecal treatment); in the case of cancer, a sample and examination of cerebrospinal fluid can be used to detect malignant cells; in the case of increased intracranial pressure due to a CNS tumour, cerebrospinal fluid removal (CSF) is also used to relieve pressure.
macrocephalus	large head, which can be caused by a hydrocephalus (hydrocephalus) in a child with unclosed fontanelles, but also by a large tumour or both
magnetic resonance imaging	diagnostic imaging method; very precise, radiation-free examination method for the visualization of structures inside the body; with the help of magnetic fields, cross-sectional images of the body are generated, which usually allow a very good assessment of the organs and many organ changes.
metastasis	1. tumour spread from the primary site of tumour to other parts of the body; characteristic feature of malignant tumours (cancer). 2. collective term for a disease process characterized by malignant cells spreading from their primary site to other areas of the body via the bloodstream and/or the lymphatic system.
metronomic therapy	use of cytostatics in relatively low doses, which has direct and indirect effects on the tumour cells and their environment; metronomic chemotherapy can, for example, target the blood vessels supplying the tumour (tumour angiogenesis) or stimulate the immune response against cancer; compared to conventional chemotherapy, metronomic chemotherapy is administered more frequently or regularly, respectively, while longer breaks between treatment blocks are waived. The goal is to not destroy the tumour, but to stabilize the tumour disease, e.g. by inhibiting the formation of new blood vessels.
molecular	at the level of molecules

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molecular genetic	referring to structure, formation, development, function and interactions of cells and cell building blocks (e.g. nucleic acids, proteins) at the molecular level; the focus is on the analysis of the genetic information stored in the nucleic acids (DNA and RNA) and its processing in the context of protein synthesis as well as gene regulation.
mutation	alteration of genetic material; it can arise without any identifiable external cause (so-called spontaneous mutation) or be caused by external influences (induced mutation). External influences include, for example, ionizing radiation or certain chemical substances (mutagens). If somatic cells are affected, it is referred to as a somatic mutation, and if germ cells are affected, it is referred to as a generative mutation. Somatic mutations are not heritable, while germ cell mutations can lead to hereditary damage. Depending on the extent of the change (single or multiple genes, larger chromosome segments or complete chromosomes), a distinction is made between point and block mutations as well as numerical and structural chromosomal aberrations.
neurological	referring to the function of the nervous system / nerve tissue
neuropsychology	subspecialty discipline of psychology that deals with the diagnosis and therapy of cognitive disorders after acquired brain damage; it deals with the functions of the brain, such as thinking (intelligence), attention, memory, language and motor skills.
neurosurgery	a branch of surgery that includes parts of the diagnosis and surgical treatment of diseases of the nervous system
Ommaya reservoir	a small plastic reservoir that can be implanted under the scalp and is then connected to one of the brain's ventricles; the shape of the Ommaya reservoir is reminiscent of a small pillow. At its bottom, it is connected by a tube (ventricular catheter) to one of the cerebral chambers (usually the right lateral ventricle) or another cavity in the brain filled with cerebrospinal fluid (CSF) (e.g. arachnoid cyst). The Ommaya reservoir (or Rickhams reservoir, another model with a similar mechanism) is implanted as part of a short, neurosurgical procedure. Such a reservoir can be connected to a shunt system for the long-term treatment of hydrocephalus or to a ventricular catheter.
palliative therapy	anti-cancer therapy that is primarily aimed at maintaining or improving the quality of life; palliative therapy becomes relevant

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	when a patient can no longer be cured. In contrast, curative therapy is primarily aimed at healing the patient.
photon	from ancient Greek light; smallest unit of electromagnetic radiation; each photon transports energy.
physical examination	an important part of diagnostic examinations; includes palpation and listening to certain body organs as well as testing reflexes to obtain indications of the nature or course of a disease.
pineal gland	hormone gland attached to the diencephalon between the two cerebral hemispheres; its function is the production of melatonin, a hormone that makes the body respond to changes in light conditions.
primary tumour	the tumour that developed first, from which metastases can originate
prognosis	prediction of the course and outcome of a disease / prospect of recovery
prognostic factors	factors that allow an approximate assessment of the further course of the disease (i.e. the prognosis);
proton	a positively charged particle within an atom; together with the electrically neutral neutrons, it forms the atomic nucleus. Protons form the counterpart to the negatively charged electrons of the atomic shell.
proton therapy	modern form of radiotherapy using protons for the treatment of malignant tumours; compared to conventional radiotherapy with photons, this type of radiation can specifically target the tumour area, thereby sparing adjacent, healthy tissue from the effects of radiation.
radiotherapy	controlled use of ionizing (high-energy) radiation for the treatment of malignant diseases
randomisation	(statistical) random distribution of patients to treatment and control groups in a study; the strict random distribution is intended to eliminate systematic errors in the evaluation of therapy studies.
remission	temporary or permanent decrease or disappearance of the signs of cancer.
Rickham reservoir	a small plastic reservoir that can be implanted under the scalp and is then connected to one of the brain's ventricles; the shape of the Rickham reservoir is reminiscent of a small pillow. At its bottom, it is connected by a tube (ventricular catheter) to one of the cerebral

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	chambers (usually the right lateral ventricle) or another cavity in the brain filled with cerebrospinal fluid (CSF) (e.g. arachnoid cyst). The Rickham reservoir (or Ommaya reservoir, another model with a similar mechanism) is implanted as part of a short, neurosurgical procedure. Such a reservoir can be connected to a shunt system for the long-term treatment of hydrocephalus or to a ventricular catheter.
seizures	uncontrolled electrical activity between nerve cells in the brain; a distinction is made between focal and generalized seizures. Focal seizures are limited to a specific area of the brain; depending on the area of the brain, the symptoms vary: e.g. twitching of one side of the body, an arm or a leg. Generalized seizures spread over large areas of the brain and lead, for example, to twitching of the limbs, sudden absence and loss of consciousness.
spinal cord	part of the central nervous system; its main function is to transmit messages between the brain and other organs of the body. The spinal cord is protectively enveloped by the three spinal cord membranes and the bony spinal canal.
supratentorial	located above the cerebellar tentorium, i.e. in the middle or anterior cranial fossa
surgery	surgical intervention on or in the body of a patient for the purpose of treatment, less often also in the context of diagnostics; the surgical intervention is carried out with the help of special instruments, generally with the patient under anesthesia.
symptom	sign of illness
syndrome	clinical presentation resulting from the coincidence of various characteristic signs (symptoms)
systemic	covering/includiing the entire body
therapy optimising trial	a controlled clinical trial (study) that aims to provide the best possible treatment for patients and at the same time to improve and develop treatment options; therapy optimisation is aimed not only at improving the chances of recovery, but also at limiting treatment-related side effects and long-term effects.
tumour	groups of abnormal cells forming a growing lump, both benign and malignant
ultrasound	an imaging technique used to examine organs, in which ultrasound waves are sent through the skin into the body; at tissue and organ boundaries, the sound waves are reflected

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	back, picked up by a receiver (transducer) and converted into corresponding images with the help of a computer.
undifferentiated	here: immature, not yet functional and usually capable of unlimited division (e.g. stem cells); the development from undifferentiated to differentiated cells and tissues (differentiation) takes place in stages. Accordingly, there are many different degrees of differentiation.
WHO classification	international guideline developed by the World Health Organization (WHO) for classification, diagnosis and differentiation/grading of (malignant) diseases

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