

Trisomy Alliance

Trisomy 13 and 18 A Family Dictionary of Medical Terms

www.internationaltrisomyalliance

A Family Dictionary of Medical Terms

by

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International Trisomy Alliance, ITA, was formed to offer parent groups, physicians and other professionals the latest research about trisomy 13 and trisomy 18, and to make available web based publications to help parents after the diagnosis, birth, or loss of a baby with one of these syndromes.

This family dictionary contains information about trisomy 13 and 18, and the language parents may encounter after a diagnosis. Helpful comments by ITA are italicized. Although medical terms are included, this is NOT a medical dictionary. ITA do not recommend or advise any particular course of action or medication, and families should always consult their doctor before changing or introducing treatments, or if a medical problem arises.

if you have an entry you would like included, please send your contribution via the contact form on the ITA website to be considered by the founders.

International Trisomy Alliance publishes several other web-based booklets with translations as available. Trisomy 13 and 18 for Younger Children and Trisomy 13 and 18 for Older Children use illustrations to explain these syndromes, Preparing for your baby's arrival is a landmark in information for prospective

parents of a baby with trisomy 13 and 18, and a **Cherished Pregnancy** looks at the practical issues when parents continue a pregnancy after a prenatal diagnosis.

The Founders of International Trisomy Alliance

Barb Farlow MBA

Barb's daughter Annie had trisomy 13 and lived for 80 days. Barb's driving goal has been to improve communication with parents and professionals.

Jenny Robbins

Her daughter Beth had trisomy 13 and lived for 3 months. Jenny co founded and was a trustee of SOFT UK for twenty years, and wrote several publications for families.

Siri Fuglem Berg MD Ph.D.

Siri is an anesthesiologist who was given a prenatal diagnosis of trisomy 18, and Evy Kristine lived for three days. Siri established a Norwegian website to support families in similar situations: www.trisomi18.com and she is a medical adviser to ITA.

Debbie Bruns Ph.D.

Debbie is the Principal Investigator of the Tracking Rare Incidence Syndromes (TRIS) Project. She worked with three preschool aged children with trisomy 18 when she was a classroom teacher.

Abnormality

Not a parent friendly term and means not normal or weirdness. It was used to describe conditions associated with trisomy 13 and trisomy 18.

Seeing a list of 'abnormalities' or 'defects' can dehumanize a baby, and in this dictionary ITA refers only to medical conditions or anomalies.

Abortion

When a pregnancy is ended before the unborn baby is capable of life outside the womb.

See Termination of pregnancy

Amniocentesis

Usually performed from 15 weeks of pregnancy onwards. A needle is inserted through the abdomen to obtain amniotic fluid with fetal cells.

See Diagnostic tests

Aniridia

The iris is the colored ring in the eye, and **aniridia** is the absence or incomplete development of one or both irises.

Apnea

The medical term for a pause in breathing lasting more than 20 seconds. About 50% of babies with trisomy 13 or 18 will have apnea, especially during early infancy. **Obstructive apnea** is when the muscles in the throat relax during sleeping and breathing can stop for more than a few seconds. A baby with **retrognathia** is more likely to experience obstructive apnea, and sleeping on the back may aggravate this condition.

Apnea is frightening for parents who have not been warned as breathing stops suddenly and a baby may become limp and blue. Breathing usually restarts after a few moments and slight stimulation of a baby, stroking their face or moving them slightly, can encourage a quick recovery unless the child is otherwise ill with pneumonia or heart failure.

Discuss with your pediatrician whether to use a **pulse oximeter** or **apnea monitor**, and the resuscitation techniques you can use if your child does not breathe after stimulation. Some parents may decide on non-intervention.

See **Pulse oximeter** and **Apnea monitor**

Apnea Monitor

A machine that monitors a baby's heartbeat and breathing, and if they slow or cease, an alarm sounds. Parents can buy a portable monitor.

Arthrogryposis

This condition means curved joints and can be mild or severe. It is often accompanied by muscle weakness that may restrict movement.

Hydrotherapy can relax stiff muscles and tendons, and physiotherapy and massage can help to make the joints suppler.

See Clubfeet

Atresia

Atresia is the absence or closure of an opening or tubular structure, for example: esophageal atresia when the tube carrying food and liquid from the mouth ends without connecting to the stomach. Infants will swallow normally but the liquid will return via the nose or mouth causing respiratory distress.

Intravenous fluids can be given when esophageal atresia is suspected. Surgery may be an option.

Birth plan

A plan made before baby's birth that takes into consideration what is known with certainty about the baby (through scans), the parents values and goals, and the options available at the hospital.

Often the plan includes many considerations for the unknown, such as the level of respiratory support needed by the baby after birth and a request for confirmation of the anomalies that were predicted to exist before birth such as brain and heart conditions.

Bronchoscopy

This is a procedure when a doctor uses an instrument called a **bronchoscope** to look into the main airways into the lungs. It is passed through the nose or mouth into the windpipe (trachea), and shines a light into the lungs to help a doctor diagnose or treat certain lung conditions and infections.

Caesarean section

A **caesarean section** is sometimes called a C-section. This is the term used when a surgical cut is made through the abdomen and womb to deliver one or more babies.

An **emergency caesarean** is performed when the life of the pregnant woman or her unborn baby is at risk, and the baby is delivered quickly.

An **elective caesarean** may be performed on medical advice if a normal vaginal delivery would increase the risk to the mother or baby.

Some doctors agree to a planned C-section if this increases the possibility of a live birth when fetal distress occurs in labor. Parents should discuss the increased risk to the mother and for later pregnancies with their doctor.

Cataract

The lens is normally clear and helps focus light on the retina at the back of the eye. A **cataract** is a clouding of the lens in one or both eyes.

Choroid plexus cysts

Fluid filled cysts called **choroid plexus cysts** may be seen in the brain of an unborn baby during an ultrasound scan, and although they are a marker for trisomy 18, many healthy babies develop them during pregnancy without ill effects.

Chorionic villus sampling

See CVS and Diagnostic tests

Chromosomes

Our body is made from cells that continue to divide as a person grows. Every cell has two pairs of chromosomes numbered 1 to 22 and each pair is different. One chromosome in each pair is inherited from the mother, and the other from the father.

The chromosomes are numbered in order of size, with chromosome number 1 being the longest. Girls also have a pair of X chromosomes, and boys have one X and one Y chromosome, making a total of 46 in each human cell. Tri means three, and trisomy means three chromosomes in each cell and not the usual pair.

See Trisomy 13 and Trisomy 18

Cleft lip and palate

A **cleft lip** is a gap in the upper lip that forms early during pregnancy. Clefts can form on other parts of the face, and many babies with this condition are otherwise healthy.

A **cleft palate** occurs when the two parts of the palate do not join properly and there is a gap in the roof of the mouth. A baby with a cleft palate cannot make a seal between the mouth and the nipple or bottle teat, and is unable to suck effectively.

There are various treatment options when a baby with a cleft lip/ palate survives infancy, and a plate can be fitted in the roof of the mouth. Many families are told their baby has a cleft after ultrasound scans, and the family can be prepared.

See Nasogastric

Colic

Colic and stomach gas are conditions that are painful but temporary, and a baby may have bouts of screaming. There is no medication that is proven to relieve the discomfort.

When a baby with trisomy 13 or 18 cries constantly the cause may be reflux for which there are medications. See **Reflux**

Coloboma

A **coloboma** is when the eyes have a missing piece of tissue and this causes a gap in the structure of the eye.

Comfort care

Comfort care usually relates to the care provided after a decision is made to forgo any treatment that might be deemed to be aggressive such as life support, (intubation, ventilation), and surgery. Care that allows a person to continue their natural life in comfort is provided, and usually includes fluids and nutrition, oxygen and antibiotics. Medication is administered as necessary to relieve discomfort or pain but not with the intent to hasten death.

ITA cautions comfort care in some institutions or countries does not include fluids and nutrition, any form of oxygen or antibiotics, and medication such as narcotics is administered in place of these comforts. Parents should not assume anything, but should ask questions and discuss fully all the options with their pediatrician before deciding the best plan for their baby.

See Palliative care

Constipation

Difficulty passing a motion or stools is common in infants with trisomy 13 or 18, and a pediatrician may advise extra fluid, medication or suppositories.

СТ

A **CT scan**, computerized tomography, is a specialized type of x-ray that uses radiation to show soft tissues that could not normally be seen on an X-ray.

Cutis aplasia

When an area of skin is missing, usually on the scalp. This is not a common condition, and the size can vary from a few millimeters upwards.

When the area is small, gentle washing and antiseptic creams can reduce the risk of infection, and this can be treated with antibiotics if it occurs.

CVS

CVS (chorionic villus sampling) or a placental biopsy can be performed between 10 and 13 weeks of pregnancy and can be performed earlier than amniocentesis. A needle may be inserted through the abdomen into the placenta and used to remove a sample of tissue, or a thin catheter is inserted through the cervix and a sample of chorionic villi cells are gently suctioned into the catheter.

See Diagnostic tests

Diagnostic tests

Genetic **amniocentesis** and **chorionic villus sampling (CVS)** are invasive diagnostic tests that identify whether an unborn baby has normal chromosomes. Ultrasound is used to guide the needle or the catheter that is used. A provisional result can be obtained within 3 days, and a full karyotype giving a more detailed result will take up to 3 weeks. Parents can be told the sex of their baby if they wish. Amniocentesis has a very small risk of causing a miscarriage, and CVS a slightly higher risk.

The exact cause of a miscarriage following an amniocentesis or CVS is unknown, and this risk continues for up to 3 weeks after the test in addition to the natural risk of a miscarriage occurring. Amniocentesis and CVS and may not be advised when a woman's previous pregnancies have ended in miscarriage, placental problems or an incompetent cervix.

See Amniocentesis and CVS

Diaphragmatic hernia

A **diaphragmatic hernia** is when there is an opening in the floor of the chest that separates the lungs from the abdomen. Contents from the abdomen may extend through this hole into the chest, thereby restricting the development of the lungs.

DNR

This is an acronym that stands for 'Do Not Resuscitate'. A DNR can mean different things, but in general it means that if the patient needs life support or chest compressions to prevent death, they are not provided. Some hospitals use the term AND which stands for 'Allow Natural Death'.

See Comfort care and Palliative care

ECG

ECG, also called an EKG, measures if the heart is functioning normally.

Small plastic patches (electrodes) connected to the ECG machine are placed on the body. A printout shows the electrical activity of the heart.

Edwards syndrome

This syndrome was named after Dr. John Edwards who first described the condition in 1960, and it is also known as trisomy 18.

Euthanasia

Euthanasia used in a medical sense means to administer medication with the intent to end the life of someone who is suffering or in pain. This is different from medication given with the intent of relieving pain or distressing medical symptoms, and which has the secondary side effect of hastening death. It is a criminal offence in most countries to give an overdose of any medication with the intent of euthanizing a baby.

In the Netherlands in 2004, the Groningen Protocol allowed the active ending of the life of an infant or child and was declared compulsory by the Dutch Society of Pediatrics in 2005. The Groningen Protocol states that only parents can start the procedure for 'expected quality of life or unbearable suffering', and the agreement of a doctor and a social worker is needed.

ITA says the protocol is flawed in many ways. On average 75 babies are euthanized each year, and the protocol has been attacked worldwide as 'an attempt to legalize infanticide'.

Failure to thrive

Babies and children with trisomy 13 and trisomy 18 will not grow as quickly, or weigh as much as a healthy baby or child of the same age.

Failure to thrive is when a baby gains weight too slowly and does not seem content.

This can be a medical problem associated with trisomy 13 or 18, and when a baby fails to grow or gain weight, supplements can be prescribed.

See Nasogastric

Feticide - medical

Medical **feticide** is when an injection is given to stop the heart of the unborn baby to avoid the risk of a live birth after the parents have agreed to terminate a pregnancy. This is legal in many countries if the unborn baby has been diagnosed with a serious condition.

In some countries however, feticide is not performed, and they encourage the mother to hold the baby after it is delivered as the baby may live for a while after the termination. Feticide is sometimes used to reduce the number of unborn babies in a multiple pregnancy.

Gastro-esophageal reflux

Gastro-esophageal reflux, or reflux, is when fluid in the stomach flows back up the food pipe into the throat (esophagus) causing an acidic burning feeling.

Chronic reflux when a baby has trisomy 13 or 18 can result in complications such as failure to thrive and pneumonia.

A doctor will be able to check whether reflux is present and may prescribe thickening for feeds or medication. Keeping the baby fairly upright immediately after a feed, giving smaller more frequent feeds, and gentle winding at shorter intervals may reduce reflux.

Gastrostomy

When a child is older they may need a feeding tube placed through an incision in the abdomen into the stomach. This is sometimes called a **G tube**, **button** or **peg**, and can provide nutrition in the longer term. Gastric feeding tubes must be fitted in hospital.

See **Nasogastric**

Geneticist

Genetics is the study of human cells. A clinical geneticist is a doctor who specializes in genetics, chromosome disorders and hereditary conditions.

Parents who wish to have another child and are concerned their next baby might have trisomy 18 or 13 should talk to a genetic counselor. The risk of recurrence is very low after having a baby with full trisomy 13 or 18.

See Chromosomes

Heart conditions

A high proportion of babies with trisomy 13 or 18 will have a heart condition. These can range from mild to life threatening, and the most common heart problems are detailed below:

- Atrial septal defect is a hole between the two upper chambers of the heart. There is often a heart murmur.
- Ventricular septal defect is a hole between the lower chambers of the heart.
- Tetralogy of fallot is a hole between the two ventricles, opening from both ventricles into the aorta, and a narrowing at or just beneath the pulmonary valve that causes cyanosis, and the term a 'blue baby'.
- Patent ductus arteriosus the ductus arteriosus usually closes soon after birth, and when a baby has trisomy 13 or 18 this may take a few weeks.

See ITA Website Published Research - Heart Surgery

Heat sensitivity

Babies with trisomy 18 can become distressed in hot weather.

Good hydration and keeping a baby cool may reduce the risk, and some parents report their children grow out of heat sensitivity.

Holoprosencephaly

Holoprosencephaly is when the forebrain fails to develop properly, and this condition can be mild or severe. A baby with this condition is more likely to have anomalies of the eyes or nose.

Research suggests that a baby who has severe holoprosencephaly (alobar or semilobar) is less likely to survive infancy.

Hydramnios

A woman may be more likely to have an excess of amniotic fluid, called **hydramnios** or **polyhydramnios** during the later stages of pregnancy when the unborn baby has trisomy 13 or 18.

Hydramnios can be treated, and a pregnancy affected by this condition should be monitored closely.

Incompatible with life

Clinical texts written decades ago state that trisomy 13 and 18 are incompatible with life.

Some babies now survive pregnancy and birth, and they can be cared for at home for months, years, and even decades in a few cases.

Kidneys

Children with trisomy 18 may be at risk of developing Wilms tumor, a tumor in the kidneys, as well as other abdominal tumors. Horseshoe kidney is when the kidneys fuse together to form a shape like a horseshoe while the baby is developing in the womb.

Children with trisomy 18 should have regular ultrasound scans in order to identify Wilms and other abdominal tumors at an early stage when they can be treatable.

Lethal

A term commonly used to describe trisomy 13 and trisomy 18 in older clinical textbooks.

Still used in genetic counseling, although it should be avoided. These syndromes can no longer be termed lethal, as 10-40% of the children will survive their first birthday.

See Incompatible with life

Microcephaly

This term describes when the head is smaller than normal

Micrognathia

This means smaller jaws, especially the lower jaw.

Micro-ophthalmus

The eyeball is smaller than is usual, and a cause poor eyesight.

Mosaic trisomy

Mosaic trisomy is when only a proportion of cells in the body have three chromosomes and the remaining cells have the usual pair. The outlook for mosaicism can be a very wide spectrum ranging from near normal to that of a full trisomy.

In some cases blood tests suggest a baby has full trisomy, however a sampling of more cells or a skin biopsy might reveal mosaicism. If a baby is doing well, parents might consider asking for additional tests for mosaicism as an incomplete diagnosis of full trisomy can affect the treatments that are offered.

Most of the clinical data refers to full trisomy 13 and 18 and may be inappropriate when applied to children with a mosaic trisomy.

Doctors do not always know how a child diagnosed with a mosaic trisomy will be affected until they grow older.

MRI

An MRI scan uses magnets and radio waves to create images, and does not use X-rays. It resembles a tunnel, and is an accurate imaging tool especially for blood vessels and soft tissues.

Nasogastric

This is when a feeding tube is placed through the nose into the stomach.

This is how most newborn babies with trisomy 13 or 18 are tube fed.

See Cleft lip, and Failure to thrive

Nasojejunal (NJ)

When a baby cannot be fed into the stomach or has severe reflux, a nasojejunal or NJ tube can be threaded through the stomach and deliver nutrition to the middle part of the small intestine. NJ tubes must be fitted in hospital.

See Cleft lip, and Failure to thrive

Naspharyngeal airway

This is a tube that is inserted through the nose when it is necessary to keep an airway open. It does not trigger a gag reflex in the baby, and has a flared end that rests against the nostril.

See **Apnea**

NICU

A NICU is a neonatal intensive care unit that will have specialist doctors called neonatologists to care for premature or very sick newborns.

Obstetrician

An obstetrician is a doctor who provides reproductive care for women during pregnancy and childbirth.

Occiput

This is the back part of the skull. The back of the head may be a little more prominent in infants with trisomy 18.

Omphalocele

It is normal for the intestines to protrude into the umbilical cord until about the tenth week of pregnancy, and then retract inside the abdomen of the unborn baby. **Omphalocele** is when the intestines remain outside the abdomen in a sac because the muscles of the abdominal wall do not develop properly, and the condition may be mild or severe.

Palliative care

A palliative care team consists of specially trained doctors, nurses and medical and non-medical professionals. Their goal is to improve quality of life physically and emotionally, relieve and soothe pain and other uncomfortable symptoms, help make difficult decisions, and provide emotional support for the whole family. Palliative care can be delivered at a hospital, an assisted living facility, or in the home.

Palliative care can be provided when a condition is chronic or life limiting, and may be given in addition to existing treatments. It can aid recovery when an illness is curable, and symptoms that can be helped include breathing problems, loss of appetite, pain, anxiety, and agitation.

Partial trisomy

Partial trisomy is when only part of the chromosome is extra. This may be caused by translocation - a rearrangement of the chromosomes where parts of one chromosome are reattached to another chromosome. It may also be caused by inversion - when a segment of a chromosome is inversed. Most of the clinical data that is available refers to full trisomy 13 and trisomy 18, and may be inappropriate when applied to children with a partial trisomy.

Patau syndrome

Patau syndrome was named after Dr Klaus Patau who first described it, and this condition is also known as **trisomy 13**.

Pediatrician

A doctor whose specialty is providing medical care for infants, children and adolescents.

Photophobia

Children with trisomy 18 often have a hypersensitivity to light and need sunglasses when in daylight.

Physiotherapy

Physiotherapy is usually therapy for limbs and muscles.

Parents can be shown how to do gentle physiotherapy as soon as a baby with trisomy 13 or 18 comes home from hospital.

Placental failure

This can be a cause of stillbirth and women should be closely monitored towards the end of the pregnancy.

Research has shown that babies delivered at full term live longer and one positive factor is a higher birth weight.

Play therapy

A baby can be helped to develop focusing and eye coordination by tracking colorful and noisy toys that are waved slowly from side to side. *Moving lights and mirrors can provide stimulation, and shiny mobiles and toys may help a baby develop the sensory skills of touch and holding.*

Polydactyly

This is when a baby sometimes has an extra finger/s or toe/s, and it may be inherited. Polydactyly is also associated with trisomy 13.

An extra digit or digits are usually smaller and can be removed when necessary.

Polyhydramnios See Hydramnios

Pre eclampsia

Pre eclampsia is the most common potentially life threatening condition that can occur during pregnancy or up to 6 weeks after delivery. If untreated it can develop into eclampsia and a baby must be delivered quickly by induction or a caesarean section.

See Caesarean section

Prophylactic

A prophylactic is a medication or treatment given as a preventative measure.

Prophylactic antibiotics may be used when a child has repeated chest or urinary tract infections to try and reduce the frequency and severity of future infections.

Pulse Oximeter

This is a non invasive way to monitor the levels of oxygen in a baby's blood, (O2 saturation) by a sensor placed on a thin part of the baby's body, often the foot.

Parents can now purchase a range of devices to monitor oxygen levels.

Retrognathia

This is when the lower jaw is behind the frontal plane of the upper jaw.

Rocker bottom feet

Rocker bottom feet are associated with trisomy 13 and 18. The heel bone is more prominent and the bottom of the foot is rounded.

Special shoes can be fitted when a child is older.

Scoliosis

This is a curvature of the spine that may develop as children grow, and there are various treatments.

Screening Tests

Pregnant women may be offered screening tests such as scans and blood tests to assess the risk of their baby having a chromosomal condition. The results can indicate if a pregnancy is in a low or high-risk category, but do not give a definite result.

An invasive **diagnostic test** such as an **amniocentesis** or **placental biopsy** such as **CVS** remains the only way at present to confirm a chromosome problem. Some couples choose not to have tests, or may ask not to be told when a problem is seen during an ultrasound scan if they would not want to consider a termination.

See, Diagnostic tests and Ultrasound screening

Seizures

When a child develops seizures they may range from occasional twitching called myoclonic jerks to severe seizures characterized by rigidity.

Medication can calm the convulsions, but it may take time to find the correct dosage.

Spina Bifida

Spina bifida is a neural tube condition when the neural tube fails to develop or close properly, and leaves part of the spinal cord exposed.

This condition can be mild or serious, and surgery may be an option.

Stillbirth

There are several definitions including: 'the birth of an infant that has died in the womb after surviving the first 20 weeks of pregnancy'.

Strabismus

This squint is a common eye condition in children with trisomy.

Syndrome

Syndrome means a group of symptoms that alert the medical profession to a condition that may be named after the person who identified it.

Some of the more common signs that alert the medical staff to suspect a chromosome condition are a light birth weight, ears set slightly lower than normal, clenched fists, unusual palm and fingertip patterns, overlapping fingers, extra digits, and slower or absent reflexes.

Termination

A procedure resulting in the destruction or expulsion of the fetus or unborn baby from the uterus or womb.

Translocation

Sometimes a baby does not have three chromosomes 13 or 18, but may have a partial trisomy. Partial trisomy is when only part of the chromosome is extra, and this again can be caused by translocation. Translocation is a rearrangement of the chromosomes, where parts of one chromosome are reattached to another chromosome. There are many different types of translocations.

Tracheomalacia

This occurs when there is a softening of the cartilage of the trachea and leads to breathing difficulties. Babies with this condition have noisy breathing, and this symptom should be investigated to determine whether treatment is necessary. Sometimes the condition improves, otherwise a tracheostomy is performed or breathing can be supported with a nasopharyngeal airway or CPAP/BIPAP, a mask delivering pressurized air.

Tracheostomy

When a surgeon makes an opening through the neck in the windpipe (trachea) to allow a baby to breathe without using their mouth or nose. The opening is usually used with a tracheostomy tube.

Translocation

Translocation is a rearrangement of the chromosomes where parts of one chromosome are reattached to another chromosome. There are many different types of translocations. Translocations may cause partial trisomy, a condition where only part of the chromosome is extra. Inheritance issues can be discussed with a clinical geneticist.

Trisomy 13

Trisomy 13 can also be called **Patau syndrome**, and there are three number 13 chromosomes in every cell. Trisomy 13 is the third most common autosomal trisomy that can result in a live birth.

See Chromosomes

Trisomy 18

Trisomy 18 can also be called **Edwards syndrome**, and there are three number 18 chromosomes in every cell. Trisomy 18 is the second most common autosomal trisomy that can result in a live birth.

See **Chromosomes**

Ultrasound scans

An ultrasound uses sound waves to produce an image, and when a medical condition is identified a doctor will then decide whether a CT or an MRI scan is needed to provide further information.

Pregnant women are usually offered ultrasound diagnostic imaging techniques called scans to check the development of the fetus. A pattern of defects called chromosomal markers, for example, **choroid plexus cysts**, **heart conditions**, clenched fists, or **rocker bottom feet** may indicate a chromosome problem, but this can only be confirmed by an invasive diagnostic test.

Nuchal translucency scans at around 11-13 weeks measure the fluid area at the back of the baby's neck, and check for the presence of nasal bone, estimate the due date, assess the baby's growth and development, and detect some neural tube defects.

Ultrasound scans at around 18-20 weeks show flat images of the internal organs, the position of the placenta and the growth of the unborn baby.

3D scans show three-dimensional external images. For example, when a cleft lip is detected, the parents are prepared before the birth and can discuss what treatments may be available when the infant grows.

4D scans produce a moving image.

Wilms Tumor

See *Kidneys*

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