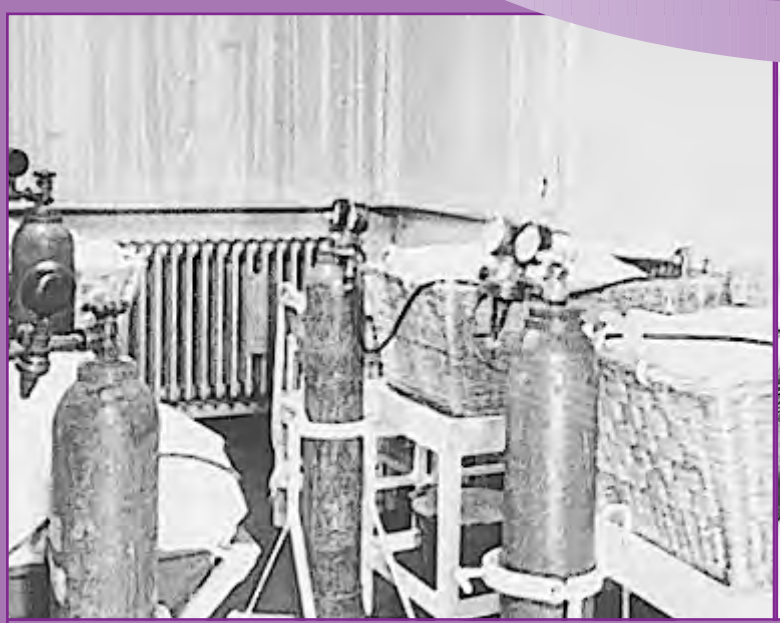
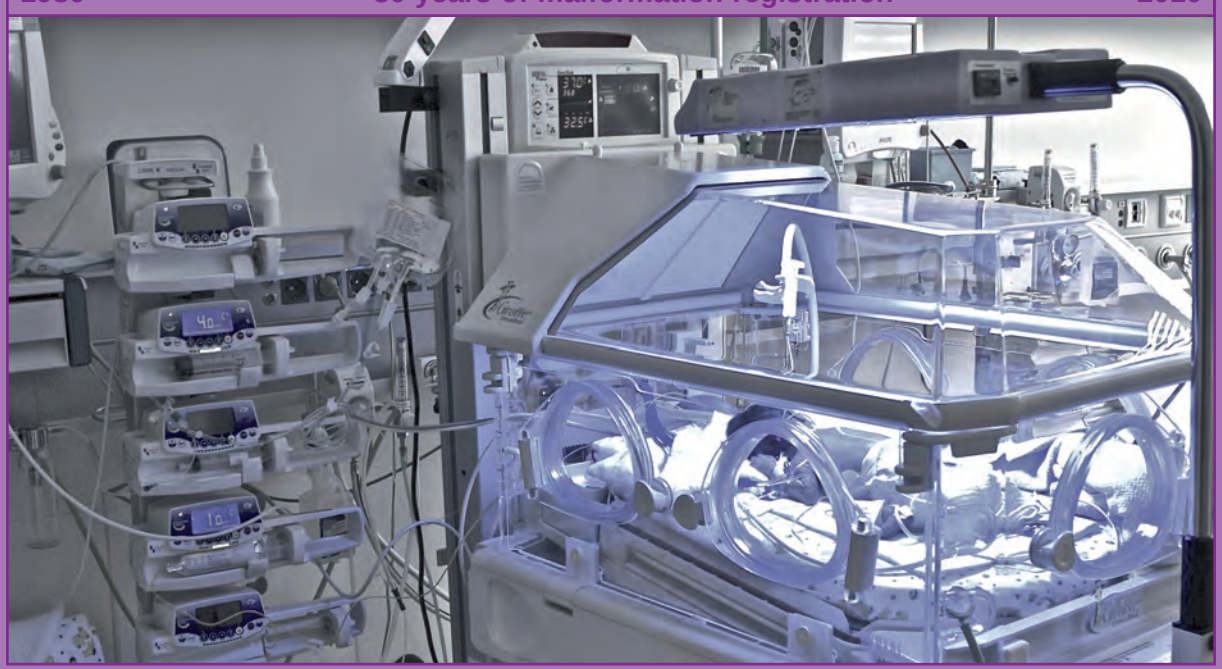
Annual Report 2010



1980 - 30 years of malformation registration - 2010



Monitoring of Congenital Malformations Saxony-Anhalt

Medical Faculty of the

Otto-von-Guericke-University Magdeburg

SACHSEN-ANHALT

Ministerium für
Arbeit und Soziales

Annual Report 2010
of the Federal State of Saxony-Anhalt
about the frequency of congenital malformations
and anomalies as well as genetically caused diseases

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Introduction

Dear reader,

Children's health condition was the main topic of the 5th Federal State Health Conference in February 2011 in Magdeburg. On this occasion the Federal State presented for the first time a children's health report. It stated that the majority of children in Saxony-Anhalt is healthy. However, healthiness and health situation still depend on the social status.

The Monitoring of Congenital Malformations Saxony-Anhalt forms an inherent part of the reporting of the general health condition in Saxony-Anhalt. They are in possession of an exemplary database which forms an excellent basis for any activity in relation to health promotion, thereby studies to prove prevention strategies can be initiated. This serves the needs to minimize risk factors and to provide specific help and consulting.

The present report not only states the development in 2010, but rather reflects 30 years of malformation surveillance in Saxony-Anhalt. To recall the history: Originally malformations were only reported in the area of Magdeburg. The reporting area was successively extended and since 2000 the whole Federal State is covered by the population-based monitoring of malformations. From the beginning of 1992 the data are analysed and in coorporation with EUROCAT (European Surveillance of Congenital Anomalies) compared to European data. Additionally, the data from Saxony-Anhalt which represents Germany is used for comparison with international malformation prevalences in ICBDSR.

The results are positive, since the infant mortality rate decreased notably during the the last ten years in Saxony-Anhalt. If we assume that the infant mortality rate is an important indicator in regard to the general evaluation of the medical attendance of pregnant women and newborns, we are happy and proud to present our results.

In comparison with all Federal States of Germany, Saxony-Anhalt occupies a top position. The steady and high quality work of the Monitoring of Congenital Malformations Saxony-Anhalt led to the achievement of the 1998 defined aim to "reduce infant mortality rate on a federal level". I would like to thank expressively all persons incorporated into the Monitoring of Congenital Malformations Saxony-Anhalt, since their work is singular in Germany. Please be reassured to continue with the same dedication to improve the child's welfare.

Your sincerely,

Norbert Bischoff
Federal Health Minister
Saxony-Anhalt

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Figures

Abbreviations

AABR	Automated auditory brainstem response
ASD	Atrial Septal Defect
bil.	Bilateral
BMI	Body-Mass-Index
BP	Basic Prevalence
CNS	Central Nervous System
dB	Decibel
DD	Differential Diagnosis
DIV	Double Inlet Ventricle
DORV	Double Outlet Right Ventricle
EUROCAT	European Surveillance of Congenital Anomalies
ENT	Ears, Nose and Throat
G-BA	Federal Joint Committee (Gemeinsamer Bundesausschuss)
ICBDSR	International Clearinghouse for Birth Defects Surveillance and Research
ICSI	Intracytoplasmatic Sperm-injection
CI	Confidence Interval
LB	Live births
MCA	Multiple Congenitale anomalies
n. o. s.	Not otherwise specified
n. s..	Not specified
NT	Nuchal Translucency
P	Prevalence
PDA	Persistend Ductus Arteriosus
PFO	Persistend Foramen Ovale
SA	Spontaneous Abortion
SB	Stillbirths
s. o.	Suspicion of
TEOAE	Transitory Evoked Otoacoustic Emissions
TOP	Termination of Pregnancy
VSD	Ventricular Septal Defect
WOG	Weeks of Gestation

1 Saxony-Anhalt - Registration Area

2 Birth Rate 2010

	Live births*	Stillbirths*	Spontaneous Abortions (> 16 WOG)	Terminations of Pregnancy	Total
Altmarkkreis Salzwedel	711	5	3	2	**721**
Anhalt-Bitterfeld	1,245	-	-	3	**1,248**
Börde	1,296	3	7	7	**1,313**
Burgenlandkreis	1,313	4	-	2	**1,319**
Dessau-Roßlau	604	3	-	3	**610**
Halle	2,185	7	3	8	**2,203**
Harz	1,518	10	-	9	**1,537**
Jerichower Land	675	2	3	2	**682**
Magdeburg	2,039	7	8	12	**2,066**
Mansfeld-Südharz	1,017	3	-	-	**1,020**
Saalekreis	1,443	4	-	2	**1,449**
Salzlandkreis	1,434	7	-	8	**1,449**
Stendal	916	5	-	8	**929**
Wittenberg	904	3	-	-	**907**
unknown district	-	-	1	1	**2**
Major cities: Dessau-Roßlau, Halle, Magdeburg	4,828	17	11	23	**4,879**
Districts, in total	12,472	46	14	44	**12,576**
Saxony-Anhalt	**17,300**	**63**	**25**	**67**	**17,455**

* Federal Statistical Office Saxony-Anhalt 2011

The total numer of 17,455 births forms the basis of the annual prevalence calculation and includes 17,300 live births, 63 stillbirths, 67 termination of pregnancy after prenatal diagnostics (all weeks of gestation) and 25 spontaneous abortions (> 16 weeks of gestation).

Our annual report refers to the births from Saxony-Anhalt, whose mother had a residence in Saxony-Anahlt during pregnancy and at point of delivery.

3 Participating Institutions of the Region 2010

3.1 Materny units / paediatric units (ordered by location)

- Klinikum Aschersleben-Staßfurt GmbH
- Klinikum Bernburg GmbH
- Gesundheitszentrum Bitterfeld/Wolfen gGmbH
- MEDIGREIF Kreiskrankenhaus Burg GmbH
- Städtisches Klinikum Dessau
- Altmark-Klinikum gGmbH Krankenhaus Gardelegen
- AMEOS Klinikum St. Salvator Halberstadt
- Sana Ohre-Klinikum GmbH Haldensleben
- Krankenhaus St. Elisabeth und St. Barbara Halle
- Universitätsklinikum Halle (Saale)
- Krankenhaus Köthen GmbH
- Klinik St. Marienstift Magdeburg
- Klinikum Magdeburg gGmbH
- Universitätsklinikum Magdeburg A.ö.R.
- Carl-von-Basedow-Klinikum Saalekreis GmbH Merseburg
- Saale-Unstrut Klinikum Naumburg
- MEDIGREIF Bördekrankenhaus gGmbH Neindorf
- Klinikum Dorothea Christiane Erxleben Quedlinburg GmbH
- Altmark-Klinikum gGmbH Krankenhaus Salzwedel
- Helios Klinik Sangerhausen
- Klinikum Schönebeck GmbH
- Johanniter-Krankenhaus Genthin-Stendal gGmbH
- Asklepios Klinik Weißenfels
- Harz-Klinikum Wernigerode-Blankenburg GmbH
- Evangelisches Krankenhaus Paul Gerhardt Stift Wittenberg
- Georgius-Agricola Klinikum Zeitz

3.2 Institutions of pre- and postnatal diagnostics (ordered by location)

- Dipl.-Med. Heweker, Fachärztin für Frauenheilkunde und Geburtshilfe, Bernburg
- Hebamme Grimm, Glindenberg/Magdeburg
- AMEOS Klinikum St. Salvator Halberstadt, Pränatale Ultraschalldiagnostik: CA Dr. Schmidt
- Dres. Perlitz, Fachärzte für Frauenheilkunde und Geburtshilfe, Haldensleben
- PD Dr. Hahmann, Facharzt für Frauenheilkunde und Geburtshilfe, Halle
- Dr. Meiner, Fachärztin für Humangenetik, Halle
- Krankenhaus St. Elisabeth und St. Barbara Halle, Pränatale Ultraschalldiagnostik: CA Dr. Seeger / OA Dr. Seliger
- Dr. Altus, Fachärztin für Humangenetik, Magdeburg
- Dr. Karstedt, Facharzt für Kinder- und Jugendmedizin, Kinderkardiologie, Magdeburg
- Dr. Karsten, Facharzt für Frauenheilkunde und Geburtshilfe, Magdeburg
- Universitätsklinkum Magdeburg A.ö.R., Institut für Humangenetik
- Universitätsklinkum Magdeburg A.ö.R., Universitätsfrauenklinik, Pränatale Ultraschalldiagnostik: OÄ Dr. Gerloff
- Universitätsklinkum Magdeburg A.ö.R., Universitätskinderklinik, Screeninglabor
- Dipl.-Med. Fiedler und Giesecke, Fachärzte für Orthopädie, Merseburg
- Dr. Schneider, Facharzt für Frauenheilkunde und Geburtshilfe, Naumburg
- Altmark-Klinikum gGmbH Krankenhaus Salzwedel, Pränatale Ultraschalldiagnostik: CA Dr. Müller
- Johanniter-Krankenhaus Genthin-Stendal gGmbH, Pränatale Ultraschalldiagnostik: CA Dr. Henschen

3.3 Pathological-anatomical institutions (ordered by location)

- Städtisches Klinikum Dessau, Institut für Pathologie
- Institut für Pathologie Dr. Taege und Dr. Bilkenroth, Eisleben
- AMEOS Klinikum St. Salvator Halberstadt, Institut für Pathologie
- Universitätsklinikum Halle (Saale), Institut für Pathologie
- Klinikum Magdeburg gGmbH, Institut für Pathologie
- Universitätsklinikum Magdeburg A.ö.R., Institut für Pathologie
- Klinikum Dorothea Christiane Erxleben Quedlinburg GmbH, Institut für Pathologie
- Praxis für Pathologie, Dr. Lüders, PD Dr. Schultz und Dr. Braxein, Stendal

4 Malformation Registration in Saxony-Anhalt

4.1 General Information

The pesent Annual Report 2010 outlines data regarding frequency of congenital malformations. At the same time, we take the opportunity to resume 30 years of malformation surveillance in our region.

The creation of a regional malformation register was purpose when in 1980 a systematical registration of congenial malformations started by the department of human genetics of the Medical Faculty Magdeburg. After the German reunification the reporting area was extended sucessively beyond the former region Magdeburg. The whole Federal State is involved into the population-based monitoring of malformations since 2000.

In 1992, the cooperation with EUOCAT (European Surveillance of Congenital Anomalies) started and since then the data was analysed and compared to other European registers. Additionally, the data from Saxony-Anhalt represent Germany within the international surveillance system of the Clearinghouse for Birth Defects (ICBDSR).

The so far registered 31 birth cohorts represent a population of 459,962 newborns. In total 46,870 anonymous data records of children and foetuses with malformations are present. The malformation rate for major malformations is at 3.3%; these malformations affect the viability of infants and usually require a postnatal intervention.

The unchanged prevalence of neural tube defects caused us in conjunction with obstetrician and neurosurgeon colleagues to present an interdisciplinary care concept for spina bifida on a prevention meeting. On this occasion, we called attention to the necessary periconceptional folic acid use again.

By presenting this Annual Report, we would like to point out also the relevance of the steady and high quality registration of malformations in our Federal State. This was only possible by the since 1995 existing funding and support of the Ministry of Employment and Social Affairs of the Federal State of Saxony-Anhalt. At this point we would like to thank especially our persons in charge in the Ministry Dr. Dr. R. Nehring, Dr. H. Willer and Dr. H. Gunkel. At the same time we would like to thank Mrs. Dipl.-Wirtsch. V. Rätzel und Dr. J. L. Hülsemann of the Medical Faculty of the Otto-von-Guericke-University Magdeburg for their competent support.

In 2010 the leadership of the Malformation Monitoring Centre Saxony-Anhalt changed. Therefore, we would like to thank again Dr. med. Pötzsch for her work.

Altogether, the singular population-based registration of malformations in Germany can only be successful in cooperation with numerous colleagues from the hospitals and ambulant institutes in Saxony-Anhalt. At this point we would like to thank everybody for participating directly or indirectly!

4.2 Registration and Analysis

The present report contains data about infants of the Federal State of Saxony-Anhalt with congenital malformations and chromosomal disorders in relation to the mother's place of residence during pregnancy, respectively at birth.

The total number of births includes
- live births,
- stillbirths,
- terminations of pregnancy after prenatal diagnostics (all weeks of gest.)
- spontaneous abortions (>16 weeks of gest.)

and forms basis for the annual prevalence calculation.

The expected date of delivery is used as the basis when analysing the termination of pregnancy, e.g. 2010 is considered the year of birth although induced termination of pregnancy took place at the end of 2009. This method is common on an international scale. Normally the time of delivery of the spontaneous abortions is not corrected.
The data of live births and stillbirths is provided by the Statistical Office of Halle.

All data transmitted to the Monitoring of Congenital Malformations are medically controlled upon receipt and the diagnoses are encoded according to ICD-10. Details about the intake of medication during the pregnancy are registered by using the internationally recommended ATC codes.

The present report outlines in chapter 7 and 8 the total number of infants with major malformations as well as the geographical distribution of appearance in the big cities and districts. Infants with only minor malformations or rather norm variations are not evaluated separately since this data is only collected incompletely in the end. The most frequent single diagnoses of major malformatios registered in 2010 are outlined in Chapter 11.

Just as in the previous years the reported pathologic prenatal screening results are analysed separately in Chapter 10.

As it is common for our reports, Chapter 12 contains the analysis of the so-called indicator birth defects registered by the ICBDSR. As we have presented data in this way for a number of years, it is possible to evaluate the current prevalences of 2010 in comparison to the last 12 years (1998-2009). A **total number** of **198,477** births forms basis for the basic **prevalence calculation**.

The grafical presentation of the annual prevalences allows to recognise frequent appearances and gives an overview about rarely appearing indicator births defects. The exact calculation of confidence levels is based on the binominal distribution with a confidence probability of 95%.

Chapter 13 of the present report outlines data regarding genetically caused diseases, chromosomal disorders, sequences, associations, complexes and embryopathies. Chapter 14 contains an analysis of malformation caused terminations of pregnancy.

As usual, chapter 16 is dedicated to a "special" topic and for the first time it is divided into two parts. On the one hand we take a look back on registration of malformations in Saxony-Anhalt since 1980. On the other hand, the anorectal malformation is treated explicitely.

The Newborn hearing screening anually forms part of the Report of the Monitoring of Congenital Malformations Saxony-Anhalt and is outlined in chapter 18.

Chapter 19 presents again the Annual Report of the department of newborn screening in Saxony-Anhalt with data of congenital metabolic disorders and endocrinopathies.

4.3 Data Quality and Completeness/Reporting Procedure

The Monitoring of Congenital Malformations Saxony-Anhalt collects data regarding malformations of newborns and foetuses since 30 years. In the beginning of the 1980s less than 200 births were registered, however, since 1997 the database increases anually by more than 2000 records. In 2010 we received information about 2313 births.

By now, we have 2149 data records for 2009; these are 100 more than we had when the Annual Report was released. The retroactively reported data which was not taken into consideration in the previous report is included in the present report and used for further evaluation.

The Monitoring of Congenital Malformations registered data of 2538 births in 2010. In 9% of all cases we received information from two or more different institutions. In this way complex diagnoses could be defined and confirmed more precisely and the constant high data quality was improved.

Also in 2010 we tried to collect for the sake of completeness the following data: month of birth (100%), birth weight (98.2%), age of the mother (99.6%) and gestational age (99.5%). These data was registered nearly complete due to the outstanding participation and commitement of all senders. We received additional information about gender (99.1%) and administrative district, repectively postal code (99.9%).

We were able to evaluate in nearly all cases of live and stilbirths (96.6%) if the infant had a normal development or if it was too small and or of low weight by regarding gestational age, birth weight and gender. We received information about the gestational age at 99.7% of all live and stillbirths. The gestational age is of importance for the classification of medical findings such as persistant foramen ovale and undescended testis.

The indication of the maternal age is of importance for chromosomal aberrations, since it is a well-known risk factor. In a few cases we received no information regarding the maternal age, however, no chromosomal aberration was registered in these cases in 2010.

The high completeness of data reflects only one side of data quality, also the number of reports we received is important when evaluating the quality. The percentage of major malformations is at 3 - 3.5% in Saxony Anhalt and Europe-wide. In administrative districts with malformation rates lower than 2%, we assume that the reporting of malformations is not well established in the corresponding clinics. Another reason might be that women residing in the administrative districts did not give birth to their children in Saxony-Anhalt.

Many malformation reports as well as information about control cases are reported by means of the **"green documentation sheets"**, which we make available to the reporting institutions free of charge. Documentation sheets may be ordered at any time by phone **+49 391-6714174** or e-mail at **monz@med.ovgu.de**. We will be at your disposal for answering any further questions about the reporting procedure and congenital malformations in general.

It is also possible to report on so-called **"white documentation sheets"**. This form serves to register a minimum data set. Here it is not necessary that parents consent to the transmission of the malformation report by signing this form.

Mostly we receive the reports by mail on our documentation form sheets. In many institutions fax reports have become the preferred method of transmission. Our fax number is: **+49 391-6714176**.
We are currently preparing an online reporting form. Please contact us if you are interested in reporting your data online.

We will be at your disposal for answering any further questions about the reporting procedure and congenital malformations in general.

6 Sex Ratio

Sex ratio of all live births and stillbirths of Saxony-Anhalt according to the information of the Statistical Office of Halle.

male	8,842	live births and stillbirths
female	8,521	live births and stillbirths
total	17,363	live births and stillbirths

sex ratio m : w = 1.04

The Statistical Office of Halle registered in 2010 a total number of 17,363 live births and stillbirths which can be split up into 17,300 live births and 63 stillbirths. Compared to the previous year (17,213 births) the total number of births in Saxony-Anhalt increased slightly (+0.9%).

The sex ratio of all live births and stillbirths shows with the value of 1.04 a light androtropism (2009: 1.07, 2008: 1.05).Similar to the previous year infants with major malformations show an androtropism with a value of 1.25 (2009: 1.28; 2008: 1.38).

Sex ratio of all births with major malformations (including abortions)

male	314	births
female	251	births
unknown	14	births
uncertain	1	birth
total	580	births

sex ratio m : w = 1.25

Sex ratio of all births with only minor malformations and anomalies

male	154	births
female	131	births
total	285	births

sex ratio m : w = 1.18

11 Organ System Involvement in Infants and Foetuses with Major Malformations

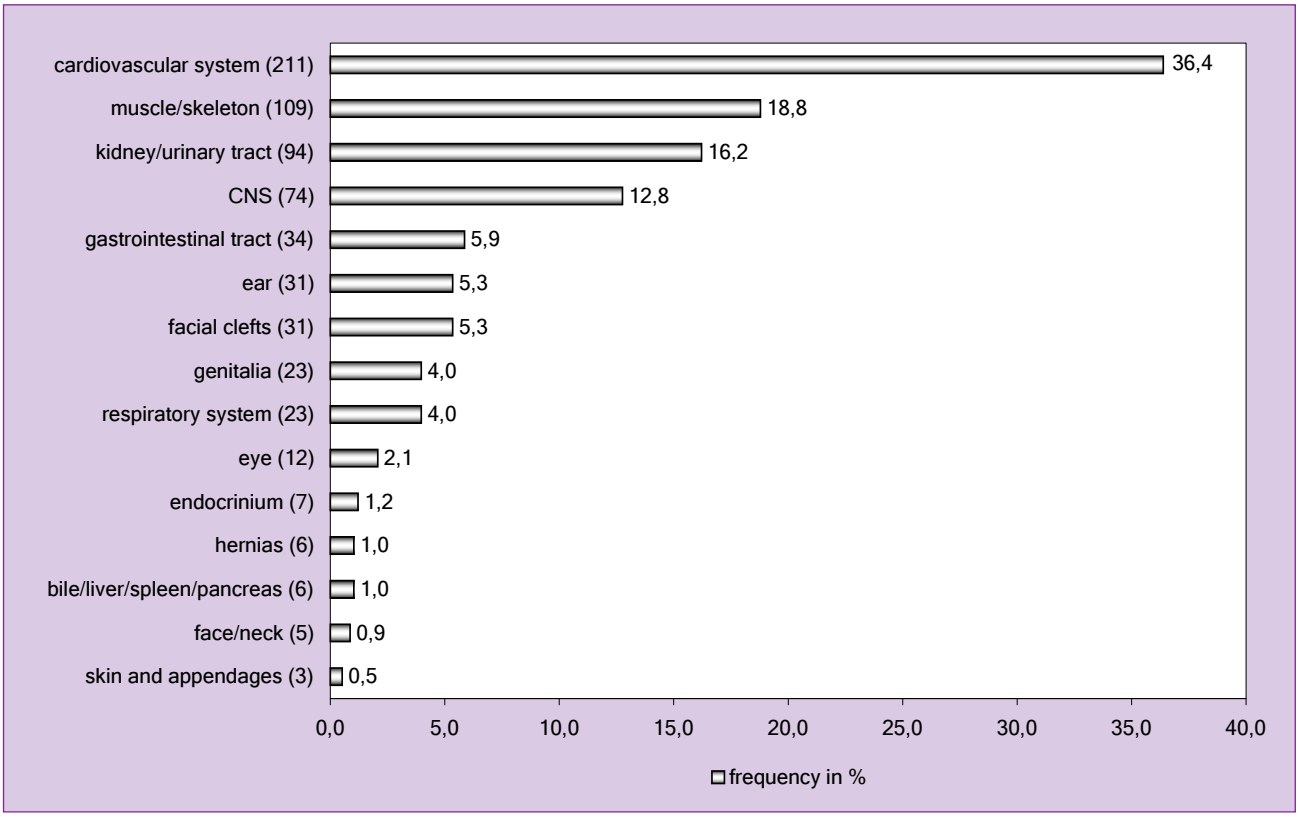


Fig. 5: Organ system involvement in major malformations (absolute figures and percentages of reported malformations)

This chapter outlines the findings of 580 infants with one or multiple major malformations. We classified the malformations according to the concerned organ system including the fact that several organ systems of an infant or foetus were concerned. Births with a MCA malformations and no indication about the single malformations are not included in the figure.

Malformations of the cardiovascular system formed the biggest part with 211 births with a diagnosis of this category and a percentage of 36.4%. This percentage lies slightly under the values of the previous years (2009: 40.6 %; 2008: 40.8 %).

The frequency in appearance of major malformations of the musculosceletal system did not change in comparison to the previous year with 18.8% (2009: 18.4 %). Malformations of the kidney and urinary tract were reported in 2010 in 16.2% of all cases, these malformations appeared more frequently in the previous years (2009: 13.8 %; 2008 12.2 %).
Malformations of the CNS were reported as one of the most frequently appearing malformation with a slightly increased percentage of 12.8% (2009: 11.1%).

Malformations of the eyes are registered rarely, however the frequency of appearance increased in 2010 (2.1%). Therefore this percentage lies above the average of the last 10 years (1.6%). The present frequency is comparable with the registered value of 2005 (2.6%).

The most frequent single diagnoses in 2010 (only major malformations)

	ICD 10	Diagnosis	Infants/Foetuses 2010		Infants/Foetuses 2000-2009
			Number	Prevalence /10.000	Prevalence /10.000
1.	Q21.1	Atrial septal defect (inclusive persistent foramen ovale/PFO)	105	60.2	68.1
2.	Q21.0	Ventricular septal defect	74	42.4	44.4
3.	Q62.3	Other obstructive defects of renal pelvis and ureter (dilated uropathy grade II-IV/ureterocele)	39	22.3	17.9
4.	Q02.	Microcephaly	36	20.6	15.7
5.	Q90.	Down's Syndrome (Trisomy 21)	33	18.9	16.3
6.	Q65.3 Q65.4 Q65.5	Congenital subluxation of hip (unilateral/bilateral/laterality unspecified)	30	17.2	17.3
7.	Q25.0	Persistant Ductus Botalli (hemodynamically effective)	29	16.6	5.9
	H90.	Perceptive of sound conduction caused hearing loss	29	16.6	5.2 (14.4*)
8.	Q69.	Polydactyly (pre- und postaxial)	27	15.5	11.1
9.	Q37.	Cleft hard and soft palate with unilateral/bilateral cleft lip	21	12.0	11.2
10.	Q66.0	Talipes equinovarus (clubfoot)	20	11.5	19.5
11.	Q62.1	Atresia and stenosis of ureter	16	9.2	6.6
12.	Q61.4	Renal dysplasia (unilateral/bilateral)/Potter II	15	8.6	5.4
13.	Q54.1 Q54.2 Q54.3 Q54.8 Q54.9	Hypospadias (without coronal/glandular)	14	8.0	7.1
14.	Q21.2	Atrial and interventricular septal defect	11	6.3	4.3
15.	Q60.0	Renal agenesia (unilateral)	10	5.7	6.9
	Q25.1	Coarctation of aorta	10	5.7	4.5
	Q42.2 Q42.3	Congenital absence, atresia and stenosis of anus with or without fistula	10	5.7	4.2
	Q91.0 Q91.1 Q91.2 Q91.3	Edward's Syndrome (Trisomy 18)	10	5.7	3.6
16.	Q30.0 Q30.1 Q30.8 Q30.9	Congenital hydrocephalus (without neural tube defect)	9	5.2	6.2

* 2007-2009 (since 2007 data collation with the newborn hearing screening tracking)

12 Indicator Defects of the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR)

12.0 Definitions

1. Neural tube defects: common congenital malformations that occur when the neural tube fails to achieve proper closure during early embryogenesis, resulting in defective development of the associated vertebral arches.
Synonyms: Spina bifida, anencephaly, NTD

2. Anencephaly: a congenital malformation characterized by the total or partial absence of the cranial vault, the covering skin, and the brain missing or reduced to small mass. Inclusive craniorachischisis. Inclusive infants with iniencephaly and other neural tube defects as Encephalocele or open spina bifida, when associated with anencephaly.Exclusive acephaly, that is, absence of head observed in amorphous acardiac twins.

3. Spina bifida: a family of congenital malformation defects in the closure of the spinal column characterized by herniation or exposure of the spinal cord and/or meges through an incompletely closed spine. Inclusive meningocele, meningomyelocele, myelocele, myelomeningocele, rachischisis. Spina bifida is not counted when present with anencephaly. Exclusive spina bifida occulta, sacrococcygeal teratoma without dysraphism.

4. Encephalocele: a congenital malformation characterized by herniation of the brain and/or meninges through a defect in the skull. Encephalocele is not counted when present with spina bifida.

5. Microcephaly: a congenitally small cranium, defined by an occipito frontal circumference (OFC) 3 standard deviation below the age and sex appropriate distribution curves.[If using a different definition or cut off point (e.g., 2 standard deviations), report but specify criteria]. Exclusive microcephaly associated with anencephaly or encephalocele

6. Congenital Hydrocephaly: a congenital malformation characterized by dilatation of the cerebral ventricles, not associated with a primary brain atrophy, with or without enlargement of the head, and diagnosed at birth. Not counted when present with encephalocele or spina bifida. Exclusive macrocephaly without dilatation of ventricular system, skull of macerated fetus, hydranencephaly, holoprosencephaly, and postnatally acquired hydrocephalus.

7. Arhinencephaly/Holoprosencephaly: a congenital malformation of the brain, characterized by various degrees of incomplete lobation of the brain hemispheres. Olfactory nerve tract may be absent. Holoprosencephaly includes cyclopia, ethmocephaly, cebocephaly, and premaxillary agenesis.

8. Anophthalmos/Microphthalmos: apparently absent or small eyes. Some normal adnexal elements and eyelids are usually present. In microphthalmia, the corneal diameter is usually less than 10 mm. and the antero posterior diameter of the globe is less than 20 mm.

9. Anotia/Microtia: a congenital malformation characterized by absent parts of the pinna (with or without atresia of the ear canal) commonly expressed in grades (I - IV) of which the extreme form (grade V) is anotia, absence of pinna. Exclusive small, normally shaped ears, imperforate auditory meatus with a normal pinna, dysplastic and low set ears.

10. Tetralogy of Fallot: a condition characterized by ventricular septal defect, overriding aorta, infundibular pulmonary stenosis, and often right ventricular hypertrophy.

11. Transposition of great vessels (TGV): a cardiac defect where the aorta exits from the right ventricle and the pulmonary artery from the left ventricle, with or without other cardiac defects. Inclusive double outlet ventricle so called corrected transposition.

12. Hypoplastic left heart sydrome: a cardiac defect with a hypoplastic left ventricle, associated with aortic and/or mitral valve atresia, with or without other cardiac defect.

13. Coarctation of the aorta: an obstruction in the descending aorta, almost invariably at the insertion of the ductus arteriosus.

14. Cleft lip with or without cleft palate : a congenital malformation characterized by partial or complete clefting of the upper lip, with or without clefting of the alveolar ridge or the hard palate. Exclusive midline cleft of upper or lower lip and oblique facial fissure (going towards the eye).

15. Cleft palate without cleft lip : a congenital malformation characterized by a closure defect of the hard and/or soft palate behind the foramen incisivum without cleft lip. Inclusive submucous cleft palate. Exclusive cleft palate with cleft lip, cleft uvula, functional short palate, and high narrow palate.

16. Choanal atresia, bilateral: congenital obstruction (membraneous or osseous) of the posterior choana or choanae. Exclusive choanal stenosis and congestion of nasal mucosa.

17. Oesophageal atresia/stenosis: a congenital malformation characterized by absence of continuity or narrowing of the esophagus, with or without tracheal fistula. Inclusive Tracheoesophageal fistula with or without mention of atresia or stenosis of oesophagus.

18. Small intestine atresia/stenosis: complete or partial occlusion of the lumen of a segment of the small intestine. It can involve a single area or multiples areas of the jejunum or ileum. Exclusive duodenal atresia.

19. Anorectal atresia/stenosia: a congenital malformation characterized by absence of continuity of the anorectal canal or of communication between rectum and anus, or narrowing of anal canal, with or without fistula to neighboring organs. Exclusive mild stenosis which does not need correction, and ectopic anus.

20. Undescended testis: bilateral undescended testes in at term newborn or at least unilateral undescended testis in males more than 1 year of age. Exclusive retractrile testis.

21. Hypospadias: a congenital malformation characterized by the opening of the urethra on the ventral side of the penis, distally to the sulcus. Incl. penile, scrotal, and perineal hypospadias. Exclusive glandular or first degree hypospadias and ambiguous genitalia (intersex or pseudohermaphroditism).

22. Epispadias a congenital malformation characterized by the opening of the urethra on the dorsal surface of the penis. Not counted when part of exstrophy of the bladder.

23. Indeterminate sex: genital ambiguity at birth that does not readily allow for phenotypic sex determination. Incl. male or female true or pseudohermaphroditism.

24. Potter sequence: a congenital malformation characterized by complete absence of kidneys bilaterally or severely dysplastic kidneys.

25. Renal agenesis, unilateral: a congenital malformation characterized by complete absence of one kidney unilaterally. Exclusive unilateral dysplastic kidney.

26. Cystic kidney: a congenital malformation characterized by multiple cysts in the kidney. Inclusive infantile polycystic kidney, multicystic kidney, other forms of cystic kidney and unspecified cystic kidney. Exclusive single kidney cyst.

27. Bladder exstrophy: complex malformation characterized by a defect in the closure of the lower abdominal wall and bladder. Bladder opens in the ventral wall of the abdomen between the umbilicus and the symphysis pubis. It is often associated with epispadias and structural anomalies of the pubic bones.

28. Polydactyly, preaxial: extra digit(s) on the radial side of the upper limb or the tibial side of the lower limb. It can affect the hand, the foot, or both.

29. Limb reduction defects: a congenital malformation characterized by total or partial absence or severe hypoplasia of skeletal structures of the limbs. Inclusive femoral hypoplasia. Exclusive mild hypoplasia with normal shape of skeletal parts, brachydactyly, finger or toe reduction directly associated with syndactyly, general skeletal dysplasia and sirenomelia.

30. Diaphragmatic hernia: a congenital malformation characterized by herniation into the thorax of abdominal contents through a defect of the diaphragm. Inclusive total absence of the diaphragm. Exclusive hiatus hernia, eventration and phrenic palsy.

31. Omphalocele: a congenital malformation characterized by herniation of abdominal contents through the umbilical insertion and covered by a membrane which may or may not be intact. Exclusive gastroschisis (para umbilical hernia), a or hypoplasia of abdominal muscles, skin covere umbilical hernia.

32. Gastroschisis: a congenital malformation characterized by visceral herniation through a right side abdominal wall defect to an intact umbilical cord and not covered by a membrane. Exclusive a or hypoplasia of abdominal muscles, skin covered umbilical hernia, omphalocele.

33. Prune belly sequence: a complex congenital malformation characterized by deficient abdominal muscle and urinary obstruction/distension. It can be caused by urethral obstruction secondary to posterior urethral valves or urethral atresia. In the affected fetus the deficiency of the abdominal muscle may not be evident. It can be associated with undescended testes, clubfoot, and limb deficiencies.

34. Down's syndrome (Trisomy 21): a congenital chromosomal malformation syndrome characterized by a well known pattern of minor and major anomalies and associated with excess chromosomal 21 material. Inclusive trisomy mosaicism and translocations of chromosome 21.

35. Patau syndrome (Trisomy 13): a congenital chromosomal malformation syndrome associated with extra chromosome 13 material. Inclusive translocation and mosaic trisomy 13.

36. Edward's syndrome (Trisomy 18): a congenital chromosomal malformation syndrome associated with extra chromosome 18 material. Inclusive translocation and mosaic trisomy 18.

Note: The prevalence rates in the following chapters are calculated population based. The value indicates the number of birth with malformations in a certain population with reference to the total number of birth in this population. Since 2000 the prevalence calculations are only referring to children whose mothers have their residence in Saxony-Anhalt. Between 1997-1999 the registration area of the Monitoring of Congenital Malformations did not cover the entire area of Saxony-Anhalt. (1997: 14, 1998: 15, 1999: 16 out of 21 administrative districts). The calculation of the basis prevalences (1998 to 2009) is based on a total number of 198,477 births.

The analysis of the indicator malformations is made with regard to the diagnoses. It is possible that one child has more than one indicator malformation. Therefore the sum of all indicator malformations would be bigger than the total number of birth with an indicator malformation.

12.1 Neural tube defects (Q00./Q01./Q05.)

	Number	Prevalence/10,000 births	Trend in comp. to basic precvalence
Major cities: 1 x Dessau-Roßlau 3 x Halle	4	8.2	↔
Districts: 1 x Burgenlandkreis 1 x Harz 1 x Mansfeld-Südharz 1 x Saalekreis 1 x Stendal 1 x unbekannt	6	4.8	↓
Saxony-Anhalt	**10**	**5.7**	↓

Neural tube defects (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence Interval (CI of 95%) /10,000 births
Cities	10.42	7.74 - 13.73
Districts	9.77	8.34 - 11.42
Region	9.93	8.66 - 11.36
EUROCAT	8.47	1.64 Spain Hospital Network* 17.73 Ukraine**

*/** Registers with lowest resp. highest prevalence/10,000 births

A total number of 10 births with neural tube defects was registered in 2010 and therefore the downward tendency of the previous years continues. The prevalence lies at 5.7 per 10,000 births and was lower than the basic prevalence of the years 1998 to 2009.

During the last 12 years a higher prevalence was determined in the major cities in than in the districts.

Compared to other EUROCAT centers our annual prevalence is within the middle range.
Compared to the published 10-years-European-trend (1999-2008) of 21 EUROCAT centers, a suprising downward tendency can be identified. This refers to anencephalus and spina bifida when the different entities of the neural tube defect are regarded isolated. The encephalocele occupies a neutral position within the trend calculation.

Compared to the basic prevalences of all EUROCAT centers for 1998 to 2009 of 8.47 per 10,000 births the value in Saxony-Anhalt of 9.93 per 10,000 births is slightly above the average.

additional Information:

Pregnancy outcome	4 x live births 6 x terminations of pregnancy
Sex	4 x male 4 x female 2 x no information
Number of isolated Malformations/MCA	4 x isolated 6 x MCA

The sex ratio of births with neural tube defects was balanced.

The most frequent entity of neural dube defects were six cases with spina bifida in 2010. Encephalocele and anencephalus occurred in two cases.

40% of infants were live births, in three cases a spina bifida occurred and in one case a minimal form of encephalocele. Only in one case of spina bifida no prenatal finding is present, in the other cases a prenatally suspected diagnosis was made. The neural tube defects have a live birth prevalence of 2.3 per 10,000 births in 2010 which ranges within the basic prevalence of live births 1998 to 2009 with 2.4 per 10,000 births.

Compared to all EUROCAT registers the basic prevalence of live births 1998 to 2009 lies with 2.66 per 10,000 births also within this range. These centres also collect data from Ireland and Malta where a termination of pregnancy after prenatal diagnostics is not allowed.

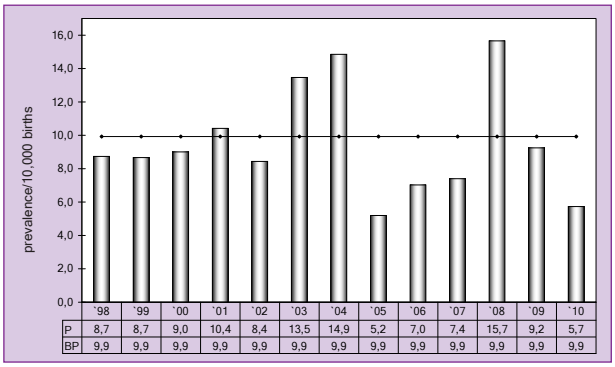


Fig. 6: Development of prevalence/10,000 births with neural tube defects in the registration area since 1998

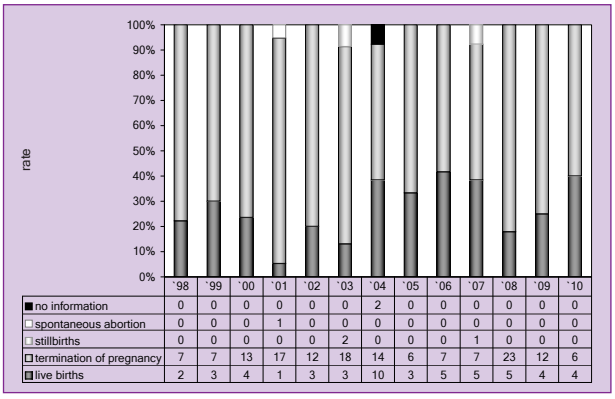


Fig. 7: Pregnancy outcomes of neural tube defects in the registration area since 1998

In 2010 one neural tube defect per 1,746 births was registered in Saxony-Anhalt.

12.2 Anencephaly (Q00.)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities	0	0.0	↓
Districts: 1 x Saalekreis 1 x unknown	2	1.6	↓
Saxony-Anhalt	**2**	**1.1**	↓

Anencephaly (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	1.67	0.72 - 3.28
Districts	2.72	1.96 - 3.70
Region	2.47	1.83 - 3.26
EUROCAT	2.85	0.24 Spain Hospital Network* 7.53 Ukraine**

*/** Centres with lowest resp. highest prevalenz/10,000 births

Two births with anencephaly were registered in 2010.

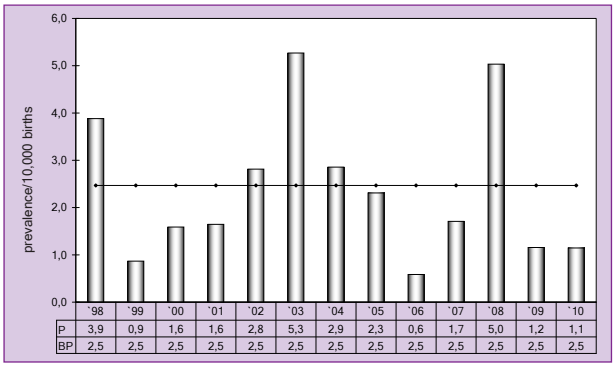
The **prevalence** of **1.1 per 10,000 births** was under the basic prevalence of the last 12 years (2.5 per 10,000 biths). The value is also in the present year below the confidence interval.

The most rare and severe form of appearence of a neural tube defect is the anencephaly. This becomes appearent also in comparison to the EUOCAT center data 1998 to 2009. The data from Saxony-Anhalt are clearly lower than the average of the single registers, since during the last 12 years the EUROCAT data (full members and asociated register) showed an average of one anencephaly per 2653 births.

additional Information:

Pregnancy outcome	2 x terminations of pregnancy
Sex	2 x no information
Number of isolated malformations/MCA	2 x isolated

The prenatally suspected diagnoses were confirmed in both cases. A confirmation by authopsy is not present in both cases. In one case we have no information where the termination of pregnancy took place. In this case a hydrops fetalis was ascertained prenatally.



	`98	`99	`00	`01	`02	`03	`04	`05	`06	`07	`08	`09	`10
P	3,9	0,9	1,6	1,6	2,8	5,3	2,9	2,3	0,6	1,7	5,0	1,2	1,1
BP	2,5	2,5	2,5	2,5	2,5	2,5	2,5	2,5	2,5	2,5	2,5	2,5	2,5

Fig. 8: Development of prevalence/10,000 births with anencephaly in the registration area since 1998

In 2010 one anencephaly per 10,000 births per 8,728 was registered in Saxony-Anhalt.

12.3 Spina bifida (Q05.)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 2 x Halle	2	4.1	↘
Districts: 1 x Burgenlandkreis 1 x Harz 1 x Mansfeld-Südharz 1 x Stendal	4	3.2	↓
Saxony-Anhalt	**6**	**3.4**	↓

Spina bifida (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	6.67	4.54 - 9.41
Districts	5.98	4.81 - 7.35
Region	6.15	5.17 - 7.29
EUROCAT	4.59	1.25 Spain Hospital Network* 10.34 Mainz (Germany)**

*/** Centres with lowest resp. highest prevalence/10,000 births

In 2010 six births with spina bifida were registered. The pevalence of 3.4 per 10,000 births is in this year under the basic prevalence of the last 12 years but within the confidence interval.

In the present year, the prevalence did not increase. However, the prevalence of live births in 2010 is with 1.7 per 10,000 slightly under the basic prevalence of the last 12 years with 2.0 per 10,000 births. Compared to the European data of the last 12 years (prevalence of 4.59 per 10,000 births) Saxony-Anhalt corresponded to the average.
Compared to the EUROCAT centres 49.6% of infants were live births. The basic prevalence of live births 1998 to 2009 of EUROCAT is at 2.3 per 10,000 live births and compared to Saxony-Anhalt with 2.0 per 10,000 live births apoximately identical.

additional Information:

Pregnancy outcome	3 x live births 3 x terminations of pregnancy
Sex	3 x male 3 x female
Number of isolated malformations/MCA	1 x isolated 5 x MCA

The sex ratio of infants/foetuses with spina bifida is balanced.

When regarding the pregnancy outcome, ratio of live births and terminations of pregnancy is balanced.

In one out of three cases of live births the malformation was not diagnosed prenatally (lumbal lesion with hydrocephaly). In the other two cases (sacral lesion without hydrocephaly and lumbal lesion with hydrocephaly) the suspected diagnosis was prenatally confirmed.

Indication about folic acid prophylaxis were made in only two cases, however none of the women already started a folic acid prophylaxis already prenatally.

The terminations of pregnancy took place after 15, 21 and 23 weeks of gestation. All three diagnoses were confirmed by authopsy.

Malformation combinations (MCA) or superordinated syndromes detected:

- Arnold-Chiari-malformation with: turricephaly
- Arnold-Chiari-malformation with: hydrocephaly and double lobed lung right
- Arnold-Chiari-malformation with: hydrocephaly, retarded hip bilateral, pes adductus bilateral
- Arnold-Chiari-malformation with: hydrocephaly, retarded hip right
- Arnold-Chiari-malformation with: imperfect triple lobar lung bilateral

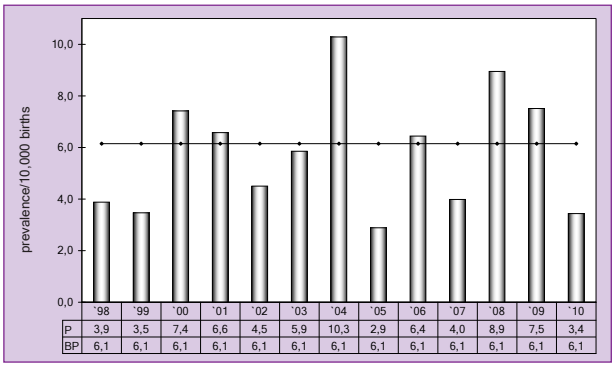


Fig. 9: Development of prevalence/10,000 births with spina bifida in the registration area since 1998

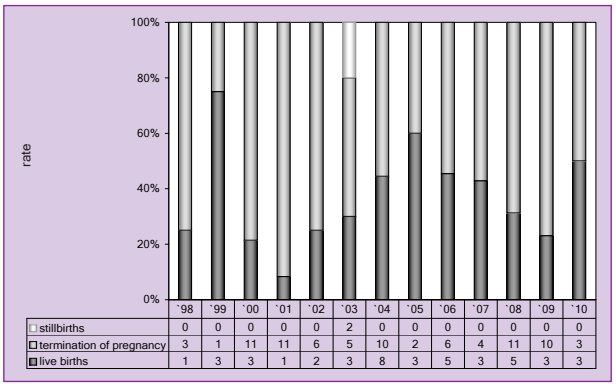


Fig. 10: Pregnancy outcomes of spina bifida in the registration area since 1998

In 2010 one spina bifida per 2909 births was registered in Saxony-Anhalt.

12.4 Encephalocele (Q01.)

	Number	Prevalence /10,000 births	Trend in comp. to basic-prevalence
Major cities: 1 x Dessau-Roßlau 1 x Halle	2	4.1	↔
Districts	0	0.0	↓
Saxony-Anhalt	**2**	**1.1**	↗

Encephalocele (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	2.71	1.44 - 4.63
Districts	1.33	0.81 - 2.05
Region	1.66	1.14 - .2.33
EUROCAT	1.05	0.19 Spain Hospital Network* 2.51 Mainz (Germany)**

*/** Centres with lowest resp. highest prevalence/10,000 births

Two births with encephalocele were registered in 2010, this is a **prevalence** of **1.1 per 10,000 births**.
The prevalence is overall small and no secure trend calculation can be made in this case.

In regard to a ten year calculation for 1999 to 2008, published by EUROCAT (data from 21 centres), the European trend is also neutral. Alltogether the basic prevalence 1998 to 2009 for Saxony-Anhalt with 1.7 per 10,000 births is within the range of all EUROCAT centre with 1.05 per 10,000 births.

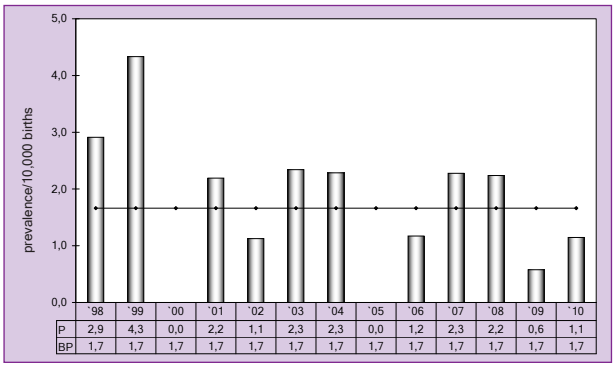
additional information:

Pregnancy outcome	1 x live births 1 x terminations of pregnancy
Sex	1 x male 1 x female
Number of isolated malformations/MCA	1 x isolated 1 x MCA

In one case the occipital encephalocele was confirmed prenatally and followed by a termination of pregnancy after 18 weeks of gestation. The diagnosis was confirmed by authopsy. We have no information about a prenatal finding in one case of a live birth with a small frontal encephalocele.

Malformation combinations (MCA) or superordinated syndromes detected:

- renal agenesia right side, hypoplasia of aorta, mesenterium, ileocolicum commune, hypoplastic gall bladder, hypoplasia of cerebellum, micropenis, dilated cerebral ventricles



	`98	`99	`00	`01	`02	`03	`04	`05	`06	`07	`08	`09	`10
P	2,9	4,3	0,0	2,2	1,1	2,3	2,3	0,0	1,2	2,3	2,2	0,6	1,1
BP	1,7	1,7	1,7	1,7	1,7	1,7	1,7	1,7	1,7	1,7	1,7	1,7	1,7

Fig. 11: Development of prevalence/10,000 births with encephalocele in the registration area since 1998

In 2010 one encephalocele per 8728 births was registered in Saxony-Anhalt.

12.5 Microcephaly (Q02.)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 1 x Halle 10 x Magdeburg	11	22.5	↔
Districts: 3 x Altmarkkreis Salzwedel 3 x Anhalt-Bitterfeld 1 x Burgenlandkreis 6 x Börde 1 x Harz 2 x Jerichower Land 1 x Mansfeld-Südharz 1 x Saalekreis 4 x Salzlandkreis 2 x Stendal 1 x Wittenberg	25	19.9	↑
Saxony-Anhalt	**36**	**20.6**	↑

Microcephaly (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	18.55	14.90 - 22.82
Districts	14.22	12.47 - 16.19
Region	15.27	13.67 - 17.04
EUROCAT	1.89	0.49 Norway* 14.64 Saxony-Anhalt (Germany)**

*/** Centres with lowest resp. highest prevalence/10,000 births

36 infants/foetuses with microcephaly were regsitered in 2010. As condition a occipito-frontal head circumference below the 3rd percentile has to be registered in regard to the gestational age.

The prevalence of 20.6 per 10,000 births is slightly higher than in the previous year and than expected.

We registered, similar to the previous years, the highest frequency in comparison to the EUROCAT data. Unfortunately, a possible non-uniform selection of criteria for a microcephaly can be the reason here.

additional information:

Pregnancy outcome	30 x live births 2 x live births descended after 7 days of life 1 x spontaneous abortion 1 x termination of pregnancy 2 x stillbirths
Sex	14 x male 22 x female
Number of isolated malformations/MCA	23 x isolated 13 x MCA

The sex ratio of microcephaly showed a clear gynaecotropy.

Six cases of microcephaly (16.7%) occured in combination with a cromosomal aberration, seven cases (19.4%) occurred in combination with at least one additional malformation. In one case a vertriculomegaly with porencephalic cysts was confirmed.

Malformation combinations (MCA) or superordinated syndromes detected:

- Edward's syndrom with: holoprosencephaly, clefting of the upper lip bilateral, VSD, hypoplasia of tentorium, craniofacial dysmorphy
- Edward's syndrome with: omphalocele, cataracta, pes equinovarus congenitus bilateral, osseous syndactyly of toes bilateral, craniofacial dysmorphy, overlapping fingers
- Down's syndrome with: unbalanced AV canal, brachycephaly, craniofacial dysmorphy, sandal's gap, single transverse palmar crease, large mammillary distance
- Down's syndrome with: ASD II, cataract bilateral
- Gorlin Goltz syndrome with: omphalocele, cleft of upper lip left, missing lower leg and foot left, osseous syndactyly and symphalangism digit 3 and 4 of the right hand, lens dislocation right side, Iris coloboma and coloboma of papilla bilateral, microcornea left side, duplex kidney, PFO at term infant, mitral valve insufficiency, retarded hip left side
- Hydrocephalus, porencephaly, Corpus callosum agenesia, retarded hip bilateral
- Gastroschisis
- Triple X (karyotyp 47,XXX)
- Fallot pentalogy, lissencephaly, malformation and hypoplasia of cerebellum, accessory finger bilateral, dysplastisc ears, PDA at preterm infant
- VSD
- penile hypospadias
- glandular hypospadias
- accessory finger left side

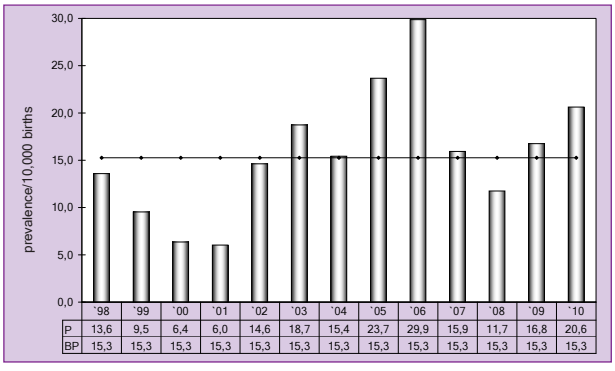


Abb. 12: Development of prevalence/10,000 births with hydrocephaly in the registration area since 1998

In 2010 one microcephalus per 485 births was registered in Saxony-Anhalt.

12.6 Congenital Hydrocephaly (Q03.)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major Cities: 1 x Halle 3 x Magdeburg	4	8.2	↔
Districts: 1 x Altmarkkreis Salzwedel 3 x Börde 1 x Mansfeld-Südharz	5	4.0	↓
Saxony-Anhalt	**9**	**5.2**	↔

Congenital Hydrocephaly (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	7.09	4.91 - .9.90
Districts-Landkreise	5.85	4.69 - 7.20
Region	6.15	5.17 - 7.29
EUROCAT	4.85	2.17 Dublin (Ireland)* 14.09 Mainz (Germany)**

*/** Centres with lowest resp. highest prevalence/10,000 births

Nine births with hydrocephalus (see inclusion criteria chapter 12.0) were registered in 2010.
The **prevalence** of **5.2 per 10,000 births** is within the basic prevalence of the years 1998 to 2009.

Compared to the European data the frequency registered in Saxony-Anhalt is within the middle range.

additional Information:

Pregnancy outcome	6 x live births 1 x live birth descended within 7 days of life 2 x terminations of pregnancy
Sex	7 x male 2 x female
Number of isolated malformations/MCA	3 x isolated 6 x MCA

Births with congenital hydrocephaly exhibit a clear androtropism.

Six cases occured in combination with additional malformations.

Malformation combinations (MCA) or superordinated syndromes detected:

- Holoprosencephaly, aplasia of the left eye, cleft of the hard palate, agenesia of the nose, macrocephaly
- Holoprosencephaly, anopthalmus bilateral, hexadactyly 5th finger right side and 5th toe bilateral, right lung incompletely triple lobed, macrocephaly, craniofacial dysmorphy, malposition of left leg
- VATER-association with: missing thumb left side, amniotic strangulation mark basal at the right thumb, VSD, inflected hands and feet, duplex spleen, fused kidney, malformed feet, macrocephaly
- Microcephaly, porencephaly, corpus callosum agenesia, retarded hip bilateral
- hypoplasia of cerebellum, megacisterna cerebellomedularis (magna), dilated cerebral ventricles, ventricle asymmetry
- Corpus callosum hypoplasia, synostosis of lambdoid suture, hypoplastic left sinus transversus

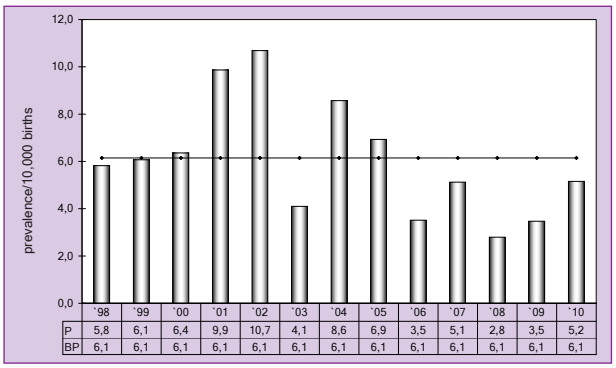


Abb. 13: Development of prevalence/10,000 births with hydrocephaly in the registration area since 1998

In 2010 one congenital hydrocephalus per 2,182 births was registered in Saxony-Anhalt.

12.7 Arhinencephaly/Holoprosencephaly (Q04.1/Q04.2)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 1 x Halle 3 x Magdeburg	4	8.2	↑
Districts: 1 x Harz 2 x Stendal	3	2.4	↑
Saxony-Anhalt	**7**	**4.0**	↑

Arhinencephaly/Holoprosencephaly (1998 to 2009)		
	Basic prevalence /10,000 births	**Confidence interval (CI of 95%) /10,000 births**
Cities	1.67	0.72 - 3.28
Districts	1.13	0.66 - 1.81
Region	1.26	0.82 - 1.86
EUROCAT	1.13	0.34 Wielkopolska (Poland)* 3.13 Mainz (Germany)*

*/** Centres with lowest resp. highest prevalence/10,000 births

Seven births with arhinencephaly/holoprosencephaly were registered in 2010. The **prevalence** lies at **4.0 per 10,000 births.**
In comparison to the previous year, this prevalence is rather high. The second highest value was registered in 2004 with 2.9 per 10,000 births.

The prevalence of 2010 in the major cities and districts is beyond the basic prevalence and beyond the confidence interval. The prevalence of this rarely appearing congenital malformation is rather high in the major cities with a value of 8.2 per 10,000 births.

Also in comparison with the European centres the arhinencephaly/holoprosencephaly appeared frequently in our registration area in 2010.

additional Information:

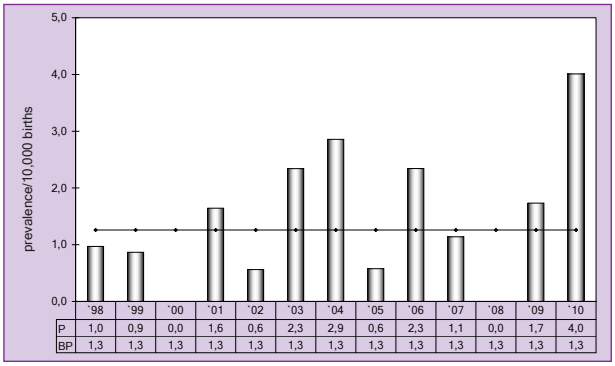
Pregnancy outcome	1 x live birth 6 x terminations of pregnancy
Sex	3 x male 2 x female 2 x no information
Number of isolierted malformations/MCA	1 x isolated 6 x MCA

Six terminations of pregnacy were carried out between 14 and 23 weeks of gestation after the diagnosis was confirmed. In one case a trisomy 18 was confirmed additionally.

One infant of twins was born alive after 35 weeks of gestation. This infant presented a lobular holoprosencephaly with hydrocephalus in addition to an aplasia of the eye, a cleft palate and an agenesia of nose.

Malformation combinations (MCA) or superordinated syndromes detected:

- Edward's syndrom with: microcephaly, cleft of upper lip bilateral, VSD, hypoplasia of tentorium, craniofacial dysmorphy
- Aplasia of the left eye, hydrocephalus, cleft of the hard palate, agenesia of the nose, macrocephaly
- Anopthalmus bilateral, hydrocephalus, hexadactyly 5th finger left side and 5th toes bilateral, right lung incompletely triple lobed, macrocephaly, craniofacial dysmorphy, malposition of legs left side
- Agenesia of the left kidney and adrenal gland, hypoplastic lung, right side unlobed , left side indicated fissura horizontalis, malformation of the craniofacial bone, flat thorax, hepatomegaly
- Hypoplastic thumb and big toe left side, pes equinovarus congenitus left side, hypoplasia of cerebellum, large base of the nose, low set ears
- Cleft lip with cleft upper jaw and palate bilateral



	`98	`99	`00	`01	`02	`03	`04	`05	`06	`07	`08	`09	`10
P	1,0	0,9	0,0	1,6	0,6	2,3	2,9	0,6	2,3	1,1	0,0	1,7	4,0
BP	1,3	1,3	1,3	1,3	1,3	1,3	1,3	1,3	1,3	1,3	1,3	1,3	1,3

Fig. 14: Development of prevalence/10,000 births with arhinencephalie/holoprosencephalie in the registration area since 1998

In 2010 one case of arhinencephalie/holoprosencephalie per 2,494 births was registered in Saxony-Anhalt.

12.8 Anophthalmos/Microphthalmos (Q11.0/Q11.1/Q11.2)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major Cities: 1 x Halle 1 x Magdeburg	2	4.1	↑
Districts: 1 x Saalekreis	1	0.8	↔
Saxony-Anhalt	**3**	**1.7**	↑

Anophthalmos/Microphthalmos (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	0.83	0.23 - 2.13
Districts	0.53	0.23 - 1.05
Region	0.60	0.31 - 1.06
EUROCAT	0.99	0.13 Zagreb (Croatia)* 3.41 Odense (Denmark)**

*/** Centres with lowest resp. highest prevalence/10,000 births

Three births with anophthalmos/microphthalmos were registered in 2010. The **prevalence** of **1.7 per 10,000 births** is in the upper range of the basic prevalence of the last twelve years. Only in 2006 a similar value was registered. The frequency of this rarely occuring malformation is not within the confidence interval.

Compared to the EUROCAT data the prevalence of 2010 occupies a position in the upper third.

additional Information:

Pregnancy outcome	1 x live birth 2 x terminations of pregnancy
Sex	2 x male 1 x female
Number of isolated malformations/MCA	3 x MCA

One live birth was registered with holoprosencephaly.

After the terminations of pregnancy the prenatally suspected diagnosis was confirmed by authopsy.

Malformation combinations (MCA) or superordinated syndromes detected:

- Trisomy 9 with: cleft lip with cleft upper jaw and palate left side incl. Cleft palate right side, renal agenesia left side , Corpus callosum agenesia, VSD, agenesia of arteria pulmonalis, right side located ascending aorta, malrotation and duplex kidney right side, multicystic extended ureter right side, hypoplastic pancreas, unlobed lung bilateral, abduction of fingers and adduction of thumbs, malformed feet, extended cranial suture, hypoglossia
- Holoprosencephaly, hydrocephalus, cleft of the hard palate, agenesia of nose, macrocephaly
- Holoprosencephaly, hydrocephalus, hexadactyly 5th finger right side and 5th toe bilateral, right lung incompletely triple lobed, macrocephaly, craniofacial dysmorphy, malposition of legs left side

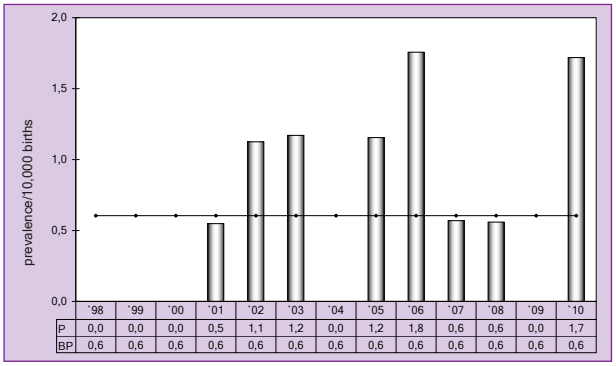


Abb. 15: Development of prevalence/10,000 births with anophthalmos/microphthalmos in the registration area since 1998

In 2010 one child with anophthalmos/microphthalmos per 5,818 births was registered in Saxony-Anhalt.

12.9 Microtia/Anotia (Q16.0/Q17.2)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 1 x Halle 1 x Magdeburg	2	4.1	↑
Districts: 1 x Börde 1 x Wittenberg	2	1.6	↔
Saxony-Anhalt	**4**	**2.3**	↗

Microtia/Anotia (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	1.46	0.59 - 3.01
Districts	1.26	0.76 - 1.97
Region	1.31	0.86 - 1.92
EUROCAT	no data available	no data available

In 2010 four births with microtia/anotia were regsitered. The **prevalence** of **2.3 per 10,000 births** is within the upper range of the basic prevalence of the years 1998 to 2009. A similar prevalence was registered in 2002. The value is slightly higher than the calculated confidence interval.

This rarely appearing malformation is not analysed by EUROCAT.

additional Information:

Pregnancy outcome	4 x live births
Sex	2 x male 2 x female
Number of isolated malformations/MCA	1 x isolated 3 x MCA

One infant with a hearing deficency bilateral and two infants with a relevant hearing defiency unilateral were registered.

Malformation combinations (MCA) or superordinated syndromes detected:

- Cleft lip with cleft upper jaw and palate right side, acoustic meatus stenosis bilateral with perceptive hearing loss (right side 70 dB, left side 50 dB), stenisos of ureteral orifice and DUP III.grade left side, plagiocephaly, ptosis right side, craniofacial dysmorphy, auricular tag right side, pelvic kidney right side
- Atresia of osseous acoustic meatus and perceptive hearing loss left side (60 dB)
- missing acoustic meatus und hearing disorder right side

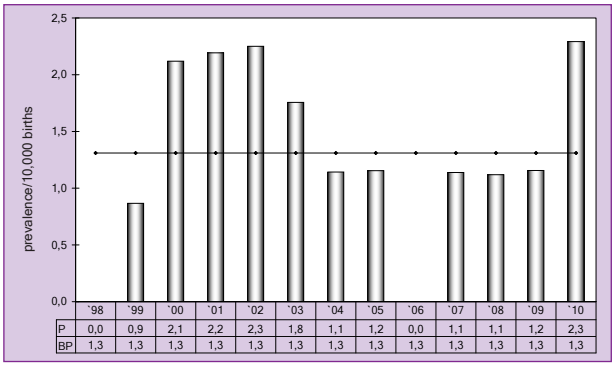


Fig. 16: Development of prevalence/10,000 births with microtia/anotia in the registration area since 1998

In 2010 one child with microtia/anotia per 4,364 births was registered in Saxony-Anhalt.

12.10 Tetralogy of Fallot (Q21.3)

	Number	Prevalence /10,000 bitrths	Trend in comp. to basic prevalence
Major cities: 1 x Halle 1 x Magdeburg	2	4.1	↔
Districts: 1 x Altmarkkreis Salzwedel 2 x Burgenlandkreis 1 x Jerichower Land	4	3.2	↔
Saxony-Anhalt	**6**	**3.4**	↔

Tetralogy of Fallot (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	3.13	1.75 - 5.16
Districts	3.32	2.47 - 4.38
Region	3.27	2.53 - 4.17
EUROCAT	2.77	0.88 Spain Hospital Network* 6.20 Northern England (UK)**

*/** Centres with lowest resp. highest prevalence/10,000 births

Six births with tetralogy of fallot were registered in 2010. The prevalence of 3.4 per 10,000 births is exactly within the expected range of the basic prevalence of the last twelve years.

In comparison with the European data the prevalence occupies a middle position within the EUOCAT centers.

additional Information:

Pregnancy outcome	5 x live births 1 x live birth descended after 7 days of life
Sex	4 x male 2 x female
Number of isolated malformations/MCA	4 x isolated 2 x MCA

All infants were live births. However, one child died within the first year of age.

With 4:2 an androtropism is present.

We received no information about the prenatal findings of these cases. Only one case occurred with non-cardial additional malformations.

Malformation combinations (MCA) or superordinated syndromes detected:

- Deletion of the short arm of chromosome 5 (Cri-du-chat syndrome) with: horseshoe kidney, DUP IV.grade right side and and III.grade left side, Pes adductus bilateral, facial hemanginoma
- ASD

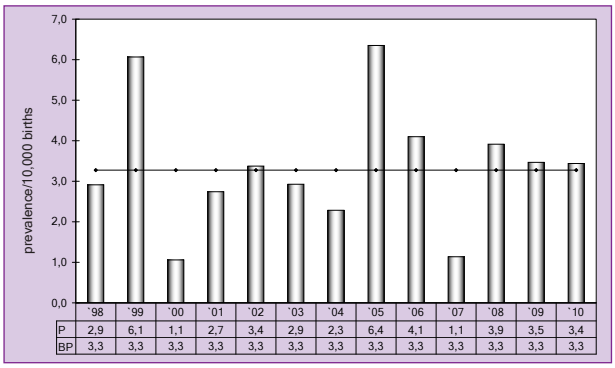


Fig. 17 Development of prevalence/10,000 births with tetralogy of Fallot in the registration area since 1998

In 2010 one tetralogy of fallot per 2,909 births was registered in Saxony-Anhalt.

12.11 Transposition of Great Vessels - TGV (Q20.1/Q20.3)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities	0	0.0	↓
Districts: 1 x Salzlandkreis	1	0.8	↓
Saxony-Anhalt	**1**	**0.6**	↓

Transposition of great vessels (1998 to 2009)		
	Basic prevalence /10,000 births	**Confidence interval (CI of 95%) /10,000 births**
Cities	6,.25	4.22 - 8.92
Districts-Landkreise	4.58	3.57 - 5.80
Region	4.99	4.05 - 6.07
EUROCAT (Q20.3)	2.86	0.80 Campania (Italy)* 5.08 Malta**

*/** Centres with lowest resp. highest prevalence/10,000 births

One live birth with transposition of great vessels was registered in 2010.
The **prevalence** of **0.6 per 10,000 births** ranges clearly below the basic prevalence of 1998 to 2009 and is outside of the confidence interval.

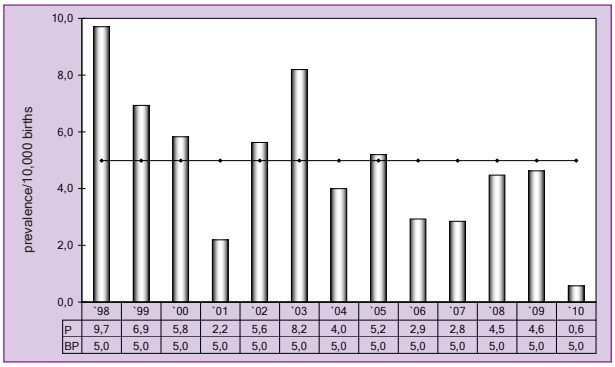
In the previous years our value occupied an upper position among the EUROCAT centres, however in 2010 the value is rather low.

additional information:

Pregnancy outcome	1 x live birth
Sex	1 x male
Number of isolated malformations/MCA	1 x MCA

Malformation combinations (MCA) or superordinated syndromes detected:

- Canalis atrioventricularis communis, supravalvular aortic stenosis, PDA at term infant (hemodynamically significant),total anomalous pulmonary venous connection, persistence of the left Vena cava superior, asplenia



	'98	'99	'00	'01	'02	'03	'04	'05	'06	'07	'08	'09	'10
P	9,7	6,9	5,8	2,2	5,6	8,2	4,0	5,2	2,9	2,8	4,5	4,6	0,6
BP	5,0	5,0	5,0	5,0	5,0	5,0	5,0	5,0	5,0	5,0	5,0	5,0	5,0

Fig. 18: Development of prevalence/10,000 births with transposition of great vessels in the registration area since 1998

In 2010 one transposition of great vessels per 17,455 births was registered in Saxony-Anhalt.

12.12 Hypoplastic Left Heart Syndrome (Q23.4)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities	0	0.0	↓
Districts: 1 x Burgenlandkreis	1	0.8	↓
Saxony-Anhalt	**1**	**0.6**	↓

Hypoplasic left heart syndrome (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	2.71	1.44 - 4.63
Districts	3.52	2.64 - 4.61
Region	3.33	2.57 - 4.23
EUROCAT	2.31	0.28 Spain Hospital Network* 4.32 Finland**

*/** Centres with lowest resp. highest prevalence/10,000 births

One birth with hypoplastic left heart syndrome was registered in 2010.
The **prevalence** of **0.6 per 10,000 births** is below the basic prevalence.

The basic prevalence of 1998 to 2009 of 3.3 per 10,000 births is compared to the EUROCAT centers within the middle range, but differs clearly from the prevalence of 2010. The prevalence of 2010 is also below the confidence inteval and even comes under the lowest value registered in 2003.

additional Information:

Pregnancy outcome	1 x live birth
Sex	1 x female
Number of isolated malformations/MCA	1 x isolated

The diagnosis was prenatally confirmed and the childbirth took place in a specialised hospital.

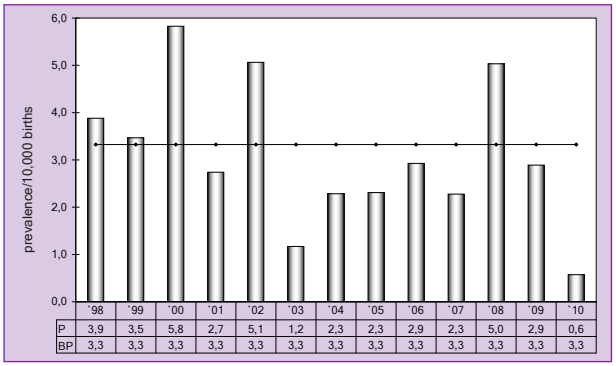


Fig. 19: Development of prevalence/10,000 births with hypoplastic left heart syndrome in the registration area since 1998

In 2010 one child with a hypolastic left heart syndrome per 17,455 births was registered in Saxony-Anhalt.

12.13 Coarctation of Aorta (Q25.1)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 3 x Halle	3	6.1	↔
Districts: 1 x Anhalt-Bitterfeld 2 x Börde 1 x Harz 1 x Jerichower Land 1 x Mansfeld-Südharz 1 x Wittenberg	7	5.6	↗
Saxony-Anhalt	**10**	**5.7**	↗

Coarctation of Aorta (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	4.79	3.04 - 7.19
Districts	4.05	3.10 - 5.21
Region	4.23	3.38 - 5.24
EUROCAT	1.46	0.14 Spain Hospital Network* 4.40 Finland**

*/** Centres with lowest resp. highest prevalence/10,000 births

In 2010 twelve births with coarctation of aorta were registered. In comparison to the previous year the value increased and the **prevalence** of **5.7 per 10,000 births** is minimally higher than the basic prevalence of 1998 to 2009.

The prevalence of 2010 is, compared to the European data, within the upper third of the single EUROCAT centres.

additional Information:

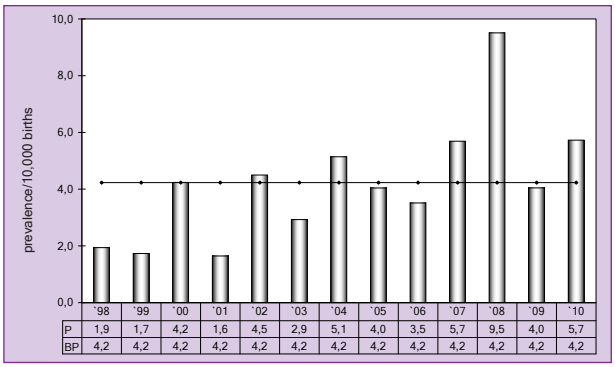
Pregnancy outcome	8 x live births 2 x terminations of pregnancy
Sex	7 x male 3 x female
Number of isolated malformations/MCA	1 x isolated 9 x MCA

In two cases a chromosomal disorder was prenatally diagnosed. In each case the termination of pregnancy took place after 8 weeks of gestation and the diagnosis was confirmed.

Non-cardial malformations occurred only at one infant with a VATER association.

Malformation combinations (MCA) or superordinated syndromes detected:

- Edward's syndrome with: cleft lip with cleft upper jaw and palate bilateral, malformed hands, pes equinovarus congenitus left side, dilatation ofarteria pulmonalis, malrotation of kidney left side, hypertelorism, pes adductus right side, overlapping finger left side
- Down's syndrome with: mesenterium ileocolicum commune, malrotation of kidneys, hypertelorism, single transverse palmar crease left side, craniofacial dysmorphy
- VATER-association with: anal atresia with fistula, thrombocyte dysfunction, malformed os sacrum, mitral valve stenosis, nondescensus testis left side and PFO at term infant, sacral dimple, small thorax, dysplastic and deep located ears, single transverse palmar crease
- Hypoplasia of aorta, VSD, ASD II, persistence of the left vena cava superior, PFO at term infant
- Hypoplasia of aorta, VSD, ASD
- VSD, ASD II, mitral valve stenosis
- VSD, ASD II
- VSD, PFO at term infant
- ASD II, PFO at term infant



	'98	'99	'00	'01	'02	'03	'04	'05	'06	'07	'08	'09	'10
P	1,9	1,7	4,2	1,6	4,5	2,9	5,1	4,0	3,5	5,7	9,5	4,0	5,7
BP	4,2	4,2	4,2	4,2	4,2	4,2	4,2	4,2	4,2	4,2	4,2	4,2	4,2

Fig. 20: Development of prevalence/10,000 births with coarctation of aorta in the registration area since 1998

In 2010 one coarctation of aorta per 1,746 births was registered in Saxony-Anhalt.

12.14 Cleft Lip With or Without Cleft Palate (Q36./Q37.)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 3 x Halle 6 x Magdeburg	9	18.4	↔
Districts: 1 x Anhalt-Bitterfeld 4 x Burgenlandkreis 1 x Börde 3 x Harz 2 x Mansfeld-Südharz 4 x Saalekreis 1 x Stendal 1 x Wittenberg	17	13.5	↔
Saxony-Anhalt	**26**	**14.9**	↔

Cleft Lip With or Without Cleft Palate (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	15.21	11.93 - 19.12
Districts	14.82	13.03 - 16.83
Region	14.91	13.34 - 16.66
EUROCAT	8.84	4.33 Spain Hospital Network* 15.94 Odense (Denmark)**

*/** Centres with lowest resp. highest prevalence/10,000 births

26 births with cleft lip with or without cleft palate were registered in 2010. The **prevalence** of **14.9 per 10,000 births** increased in comparison to the last year and correponds exactly to the basic prevalence of the years 1998 to 2009.

Compared to the EUROCAT data our value is within the upper middle range.

Six cases of this oriofacial fissure formation occured in combination with a chromosomal disorder. The terminations of pregnancy took place between 14 and 22 weeks of gestation when additional malformations were confirmed or in case of the prenatally confirmed trisomy 18 and 9, a complex vitium and a holoprosencephaly.

additional Information:

Pregnacy outcome	19 x live births 1 x spontaneous abortion 6 x terminations of pregnancy
Sex	12 x male 12 x female 2 x not specified
Number of isolated malformations/MCA	11 x isolated 15 x MCA

17 cases of cleft lip with or without cleft palate, five cases of cleft lip and 4 cases of cleft lip and upper jaw were registered. In three cases the cleft lip with cleft upper jaw and palate included a cleft of the hard palate unilateral.

The oriofacial fissure formation was confirmed in nine cases on the right side, in nine cases on the left side and in 5 cases bilateral. In three cases we received no information about the lateral occurrence.

Malformation combinations (MCA) or superordinated syndromes detected::

- Trisomy 9 with: microphthalmus bilateral, renal agenesis left side, corpus callosum agenesia, VSD, agenesie of arteria pulmonalis, right side locatedaorta ascendens, malrotation und duplex kidney right side, multicystic extended ureter right side, hypoplastic pancreas, unlobed lung bilateral, abduction of fingers and adduction of thumbs, malformed feet, large cranial suture, hypoglossia
- Edward's syndrome with: Holoprosencephaly, microcephaly, VSD, hypoplasia of tentorium, craniofacial dysmorphy
- Edward's syndrome with: Coarctation of aorta, malformed hands, pes equinovarus congenitus left side, dilatation of arteria pulmonalis, malrotation of right kidney, hypertelorism, pes adductus right side, overlapping fingers left
- Edward's syndrom with: pes equinovarus congenitus blt.
- Gorlin-Goltz syndrom with: omphalocele, microcephaly, missing lower leg and foot left side, osseous syndactyly and synphalangy digit 3 and 4 of the right hand, dislocation of lens right side, iris coloboma and coloboma of the papilla bilateral, microcornea left side, duplex kidney bilateral, PFO, mitral valve insufficiency, retarded hip left side
- Holoprosencephaly
- Microtia right side, stenosis of acoustic meatus bilateral with perceptive hearing loss (right side 70 dB, left side 50 dB), stenisos of ureteral orifice and DUP III.grade left side, plagiocephaly, ptosis right side, craniofacial dysmorphy, auricular tag right side, pelvic kidney right side
- Ebstein's anomaly, malformation of the ears with hearing disorder bilateral, malformation of the papilla left side, lobed kidney left side
- Complex cardiac defect, multicystic kidney, craniofa cal dysmorphy
- 2 x ASDII (1 x with PDA at term infant)
- Excess marker chromosome (karyotype 47,XX,+mar(15))
- Mesenterium ileocolicum commune, hypertelorism, low set ears
- Hearing loss due to perceptive hearing loss bilateral (right 40 dB, left 50 dB)
- Hip subluxation

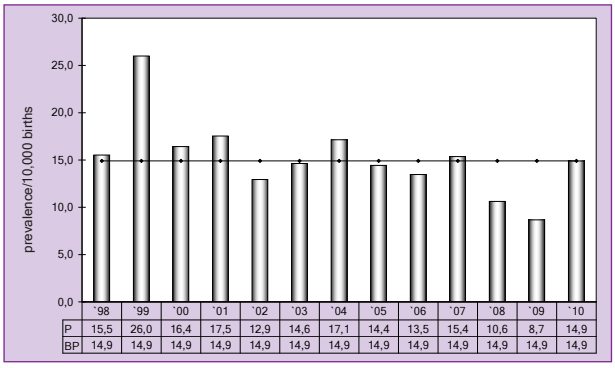


Fig. 21: Development of prevalence/10,000 births with cleft lip with or without cleft palate in the registrationarea since 1998

In 2010 one child with cleft lip with or without cleft palate per 671 births was registered in Saxony-Anhalt.

12.15 Cleft Palate (Q35.1/Q35.3/Q35.5/Q35.9)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 1 x Dessau-Roßlau 1 x Halle 1 x Magdeburg	3	6.1	↔
Districts: 1 x Anhalt-Bitterfeld 1 x Jerichower Land 1 x Saalekreis 1 x Salzlandkreis 1 x Stendal	5	4.0	↓
Sachsen-Anhalt	**8**	**4.6**	↓

Cleft Palate (1998 to 2009)		
	Basic prevalence /10,000 births	**Confidence interval (CI of 95%) /10,000 births**
Cities	7.29	5.08 - 10.14
Districts	8.57	7.25 - 10.12
Region	8.26	7.12 - 9.58
EUROCAT	6.10	3.02 Barcelona (Spain)* 13.92 Finland**

*/** Centres with lowest resp. highest prevalence/10,000 births

In 2010 eight births with cleft palate were registered in Saxony-Anhalt. The **prevalence** of **4.6 per 10,000 births** slightly decreased in comparison to the previous year.

In comparison to the basic prevalence of the last twelve years, our prevalence is clearly under the expected value.

In 2010 we did not register a similar high prevalence as in the previous years. Compared to the EUROCAT data the value is within the middle range.

additional Information:

Pregnancy outcome	8 x live births
Sex	4 x male 4 x female
Number of isolated malformations/MCA	6 x isolated 2 x MCA

All infants were live births. The sex ratio is balanced.

Malformation combinations (MCA) or superordinated syndromes detected:

- Holoprosencephaly, aplasia of the left eye, hydrocephalus, agenesie of the nose, macrocephaly
- accessory 6th finger bilateral

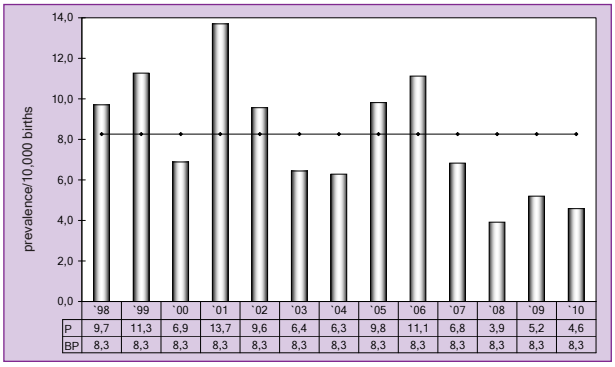


Fig. 22: Development of prevalence/10,000 births with cleft palate in the registration area since 1998

In 2010 one child with cleft palate per 2,182 births was registered in Saxony-Anhalt.

12.16 Choanal Atresia (Q30.0)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 1 x Magdeburg	1	2.0	↑
Districts: 1 x Burgenlandkreis	1	0.8	↔
Saxony-Anhalt	**2**	**1.1**	↑

Choanal Atresia (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	0.21	0.01 - 1.16
Districts	0.40	0.15 - 0.87
Region	0.35	0.14 - 0.73
EUROCAT	0.68	0.30 Campania (Italy)* 2.82 Mainz (Germany)**

*/** Centres with lowest resp. highest prevalence/10,000 births

In the last four years no choanal atresia was registered, however in 2010 two births with this diagnosis were registered. Therefore the **prevalence** is **1.1 per 10,000 births**.

This value is, similar to the basic prevalence of the last twelve years outside the confidence interval.

Within the period of 1998-2009 only nine births with choanal atresia were registered.

additional Information:

Pregnancy outcome	2 x live births
Sex	1 x male 1 x female
Number of isolated malformations/MCA	2 x isolated

Both infants were live births.

In both cases the choanal atresia occurred unilateral.

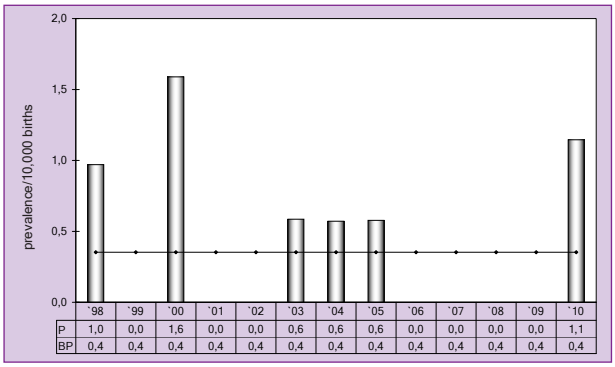


Fig. 23: Development of prevalence/10,000 births with choanal atresia in the registration area since 1998

In 2010 one child with a choanal atresia was registered per 8,728 births in Saxony-Anhalt.

12.17 Oesophageal Atresia/-Stenosis/-Fistula (Q39.0-Q39.4)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 1 x Dessau-Roßlau 2 x Halle	3	6.1	↔
Districts: 1 x Anhalt-Bitterfeld 1 x Stendal	2	1.6	↔
Saxony-Anhalt	**5**	**2.9**	↔

Oesophageal Atresia/-Stenosis/-Fistula (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	3.96	2.38 - 6.18
Districts	2.19	1.51 - 3.08
Region	2.62	1.96 - 3.44
EUROCAT (Q39.0-Q39.1)	2.24	0.97 SE Ireland* 4.33 Odense (Denmark)**

*/** Centres with lowest resp. highest prevalence/10,000 births

Five births with oesophageal atresia were registered in 2010. The **prevalence** shows with **2.9 per 10,000 births** a constant level.
Therefore the value is within the expected range of the basic prevalence of the last twelve years.

According to the by literature described distribution of frequency, in four cases the oesophageal atresia occured in combination with oesophago tracheal fistula (type IIIb According to Vogt). In one case the fistula was not reported.

additional Information:

Pregnancy outcome	3 x live births 1 x termination of pregnancy 1 x stillbirth
Sex	4 x male 1 x female
Number of isolated malformations/MCA	1 x isolated 4 x MCA

The prevalence shows an androtropism.

According to the by literature described distribution of frequency, in four cases the oesophageal atresia occurred in

Malformation combinations (MCA) or superordinated syndromes detected:

- Edward's syndrome with: omphalocele, canalis atrioventricularis communis, persistence of the left vena cava superior, hypoplasia of aorta, Horseshoe kidney, overlay of the bowed 3rd and 4th finger by the 2nd and 5th finger bilateral, low set ears
- VATER-association with: analatresia with fistula, duplex left kidney, tethered cord with lipom, malformed os sacrum, VSD, PFO and PDA at term infant
- Aanal atresia, missing testes left side and nondescensus testis right side at term infant, additional groove in the left upper pulmonary lobe
- CHARGE-association with: vascular ring of the right subclavicular artery, coloboma of the opticus, of the retina and of the choroidea bilateral, tracheomalacia, hearing loss by perceptive hearing loss bilateral (left side 70 dB, right side 40 dB), VSD, ASD I, ASD II, PDA at term infant, retarded hip bilateral

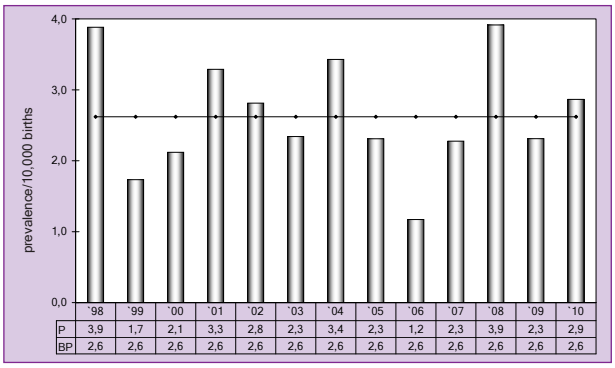


Fig. 24: Development of prevalence/10,000 births with oesophageal atresia/stenosis/fistula in the registration area since 1998

In 2010 one oesophageal atresia/fistula per 3,491 births was registered in Saxony-Anhalt.

12.18 Small Intestine Atresia/Stenosis (Q41.1/Q41.2/Q41.8/Q41.9)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 1 x Halle	1	2.0	↔
Districts	0	0.0	↓
Saxony-Anhalt	**1**	**0.6**	↓

Small Intestine Atresia/Stenosis (1998 to 2009)		
	Basic prevalence /10,000 births	**Confidence interval (CI of 95%) /10,000 births**
Cities	1.25	0.46 - 2.72
Districts	2.26	1.56 - 3.16
Region	2.02	1.44 - 2.74
EUROCAT (Q41.1-Q41.8)	0.70	0.11 Poland* 1.95 Isle de la Reunion (France)**

*/** Centres with lowest resp. highest prevalence/10,000 births

In 2010 the regressive trend of this malformation was confirmed and only one birth with small intestine atresia was registered. The **prevalence** shows a value of **0.6 per 10,000 births**.

The prevalence shows a value of 0.6 per 10,000 births. This prevalence is below the basic prevalence of 1998 to 2009 with 2.02 per 10,000 births and also below the confidence interval.

Compared with the European data of the EUROCAT centres the prevalence of 2010 is within the middle range.

additional Information:

Pregnancy outcome	1 x live birth
Sex	1 x male
Number of isolated malformations/MCA	1 x isolated

The infant was born alive at prenatally suspected a small intestine atresia.

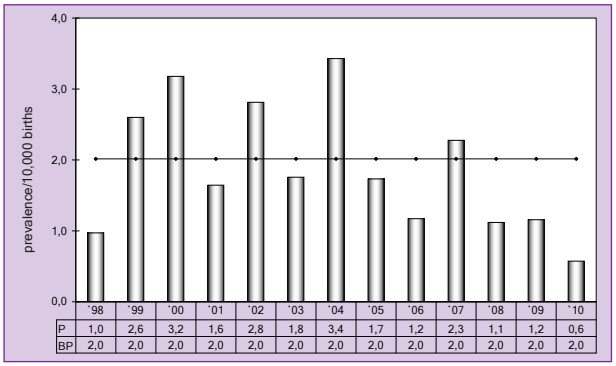


Fig. 25: Development of prevalence/10,000 births with small intestine atresia/stenosis in the registration area since 1998

In 2010 one small intestine atresia/stenosis per 17,455 births was registered in Saxony-Anhalt.

12.19 Anorectal Atresia/Stenosis (Q42.0-Q42.3)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 1 x Dessau-Roßlau 1 x Halle 2 x Magdeburg	4	8.2	↑
Districts: 1 x Altmarkkreis Salzwedel 1 x Anhalt-Bitterfeld 2 x Mansfeld-Südharz 3 x Salzlandkreis	7	5.6	↔
Saxony-Anhalt	**11**	**6.3**	↑

Anorectal Atresia/Stenosis (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	4.79	3.04 - 7.19
Districts	4.45	3.45 - 5.65
Region	4.53	3.65 - 5.57
EUROCAT	2.95	1.35 S Portugal* 7.16 Styria (Austria)**

*/** Centres with lowest resp. highest prevalence/10,000 births

In 2010, eleven births with anorectal atresia/stenosis were registered. Compared to the previous years the **prevalence** of this malformation decreased and showed 2010 a value of **6.3 per 10,000 births**.

In regard to the prevalence course an increasing trend can be observed. Chapter 16.2 outlines a detailed analysis of the data registered from 1994 until 2010.

Compared to the EUROCAT centres our prevalence is again within the upper third.

additional Information:

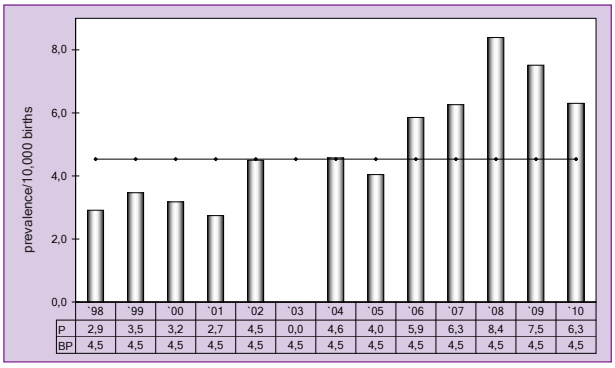
Pregnancy outcome	9 x live births 1 x live births descended after 7 days of life 1 x stillbirth
Sex	5 x male 6 x female
Number of isolated malformations/MCA	4 x isolated 7 x MCA

In 2010 a slight gynaecotropism could be observed. Since 1994 109 cases of anorectal atresia were registerd, in 56 cases (51.4 %) a male gender was confirmed.

In 2010 ten births with anorectal atresia were registered, six cases occurred in comparison with a fistula. In one case a rectum stenosis was confirmed at a preterm infant. In three cases an isolated rectum or anal atresia / stenosis was diagnosed. A chromosomal disorder was confirmed in one case.
Since the frequency of appearance increased since 1994 we participate in the german-wide network of congenital uro-rectal malformations (CURE-Net).

Malformation combinations (MCA) or superordinated syndromes detected:

- Down' s syndrom with: VSD, atrophy of the optic nerve bilateral, nondescensus testis at term infant bilateral, ventricular asymmetry, craniofacial dysmorphie, bilateral, pes adductus , sandal's gap and single transverse palmar crease
- VATER-association with: coarctation of aorta, thrombocyte dysfunction, malformed os sacrum, mitral valve stenosis, nondescensus testis left side and PFO at term infant, sacral dimple, small thorax, dysplastic and low set ears, single transverse palmar crease
- VATER-association with: oesophageal atresia with fistula, duplex kidney left side, tethered cord with lipoma, malformed os sacrum, VSD, PFO and PDA at term infant
- oesophageal atresia with: fistula, missing testes left side and nondescensus testis right side at term infant, additional groove in the left lung lobe
- 2 x PFO at term infant (1 x with glandular hypospadias)
- PDA at preterm infant (hemodynamical significant), large cavum septum pellucidum, prominent clitoris



	`98	`99	`00	`01	`02	`03	`04	`05	`06	`07	`08	`09	`10
P	2,9	3,5	3,2	2,7	4,5	0,0	4,6	4,0	5,9	6,3	8,4	7,5	6,3
BP	4,5	4,5	4,5	4,5	4,5	4,5	4,5	4,5	4,5	4,5	4,5	4,5	4,5

Fig. 26: Development of prevalence/10,000 births with anorectal atresia/-stenosis in the registration area since 1998

In 2010 one anorectal atresia/ stenosis per 1,587 births was registered in Saxony-Anhalt.

12.20 Undescended Testis (Q53.1-Q53.9)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 2 x Halle 2 x Magdeburg	4	8.2	↓
Districts: 1 x Burgenlandkreis 1 x Jerichower Land 1 x Mansfeld-Südharz 1 x Salzlandkreis 1 x Wittenberg	5	4.0	↓
Saxony-Anhalt	**9**	**5.2**	↓

Undescended Testis (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	17.09	13.60 - 21.21
Districts	6.31	5.11 - 7.72
Region	8.92	7.72 - 10.29
EUROCAT	no data available	no data available

In 2010 nine births with undescended testis at term infant were regsitered. Therefore, the **prevalence** of **5.2 per 10,000 biths** is below the basic prevalence and not within the confidence interval in 2010.

No EUROCAT data for comparison is present for undescended testis.

In three cases the undescended testis occurred bilateral. In the remaining six cases the undescended testes occurred five times on the right side. In all cases at least one additional small malformation was registered.

additional Information:

Pregnancy outcome	7 x live births 1 x live birth descended after 7 days of life 1 x stillbirth
Sex	9 x male
Number of isolated malformations/MCA	4 x isolated 5 x MCA

Malformation combinations (MCA) or superordinated syndromes detected:

- Down's syndrome with: anal atresia, VSD, atrophy of the optic nerve bilateral, ventricular asymmetry, craniofacial dysmorphy, bilateral, pes adductus, sandal' s gap and single transverse palmar crease
- VATER-association with: anal atresia with fistula, coarctation of aorta, thrombocyte dysfunction, malformed os sacrum, mitral valve stenosis, PFO at term infant, sacral dimple, small thorax, dysplastic and low set ears, single transverse palmar crease
- Oesophagus atresia with fistula, anal atresia, missing testes left side, additional groove in the left upper lobe of the lung
- Adams-Oliver-syndrome with: ectrodactyly, truncus arteriosus communis, VSD, aortic valve insufficiency, ASD II, hemangioma
- Hypospadia, auricular tag bilateral

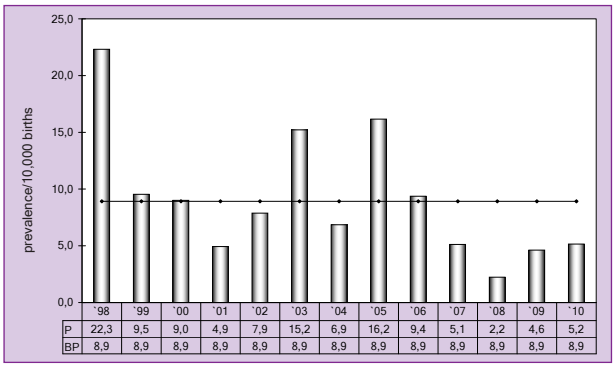


Fig. 27: Development of prevalence/10,000 births with undescended testis in the registration area since 1998

In 2010 one child with undescended testis per 1,939 births (988 boys) was registered in Saxony-Anhalt.

12.21 Hypospadias (Q54.0-Q54.3/Q54.8/Q54.9)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 1 x Dessau-Roßlau 8 x Halle 5 x Magdeburg	14	28.7	↑
Districts: 3 x Altmarkkreis Salzwedel 1 x Anhalt-Bitterfeld 2 x Burgenlandkreis 1 x Börde 4 x Harz 1 x Jerichower Land 9 x Mansfeld-Südharz 1 x Saalekreis 3 x Salzlandkreis	25	19.9	↔
Saxony-Anhalt	**39**	**22.3**	↔

Hypospadias (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	19.80	16.03 - 24.19
Districts	17.87	15.90 - 20.08
Region	18.34	16.58 - 20.27
EUROCAT	13.95	1.87 Spain Hospital Network* 34.78 Malta**

*/** Centres with lowest resp. highest prevalence/10,000 births

39 births with hypospadias were registered in 2010. The prevalence is similar to the previous years at 22.3 per 10,000 births. We registered cases of different severety.

In comparison to the EUROCAT data, our prevalence is also in 2010 in the upper third.

24 boys showed a glandular or coronar hypospadias, two boys showed a penoscrotal hypospadias. In twelve cases we were not informed about the classification of the hypospadias.

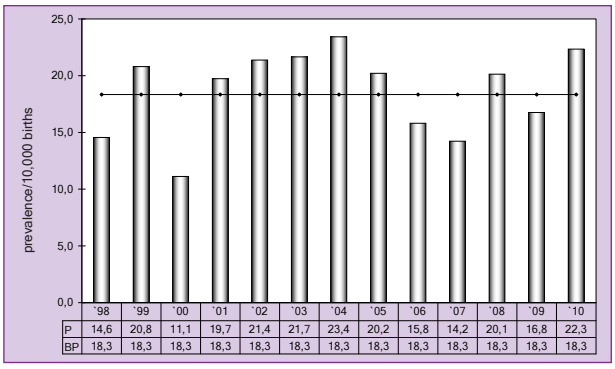
additional Information:

Pregnancy outcome	38 x live births 1 x termination of pregnancy
Sex	39 x male
Number of isolated malformations/MCA	27 x isolated 12 x MCA

In one case the hypospadias formed part of a complex malformation syndrome which was prenatally diagnosed. The termination of pregnancy took place after 23 weeks of gestation in this case.

Malformation combinations (MCA) or superordinated syndromes detected:

- Pallister-Killian syndrom with: canalis atrioventricularis communis, mesenterium ileocolicum commune, caecum mobilae, unlobed lung, rocker bottom foot, hexadactyly at the right foot, postaxial polydactyly and membranous syndactyly at the left hand, scrotum bipartum, micropenis, craniofacial dysmorphy, low set and hypoplastic finger and toe nails, shortened humerus, cystic adrenal gland right side
- 2 x microcephaly
- Anal atresia, PFO at term infant
- Nondescensus testis at term infant bilateral, auricular tag bilateral
- ASD, PFO at term infant, aortic valve stenosis
- ASD
- DUP II.grade right side
- Hernia inguinalis right side, auricular fistula left side, preauricular tag right side
- Accessory urethra, cleft preputium, retarded hip bilateral
- 2 x lateral penile curvature (1 x with preputial apron)



	'98	'99	'00	'01	'02	'03	'04	'05	'06	'07	'08	'09	'10
P	14,6	20,8	11,1	19,7	21,4	21,7	23,4	20,2	15,8	14,2	20,1	16,8	22,3
BP	18,3	18,3	18,3	18,3	18,3	18,3	18,3	18,3	18,3	18,3	18,3	18,3	18,3

Fig. 28: Development of prevalence/10,000 births with hypospadias in the registration area since 1998

In 2010 one hypospadias per 448 births (228 boys) was found in Saxony-Anhalt.

12.22 Epispadias (Q64.0)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities	0	0.0	↓
Districts: 1 x Saalekreis	1	0.8	↔
Saxony-Anhalt	**1**	**0.6**	↔

Epispadias (1998 to 2009)		
	Basic prevalence /10,000 births	**Confidence interval (CI of 95%) /10,000 births**
Cities	0.21	0.01 - 1.16
Districts	0.40	0.15 - 0.87
Region	0.35	0.14 - 0.73
EUROCAT	no data available	no data available

In 2010 one birth with the very rarely appearing malformation epispadias was registered in Saxony-Anhalt. The **prevalence** is at **0.6 per 10,000 births**. Regarding the basic prevalence of the years 1998 to 2009 the value is within the expected range.

additional Information:

Pregnancy outcome	1 x live birth
Sex	1 x male
Number of isolated malformations/MCA	1 x MCA

Malformation combinations (MCA) or superordinated syndromes detected:

- ASD

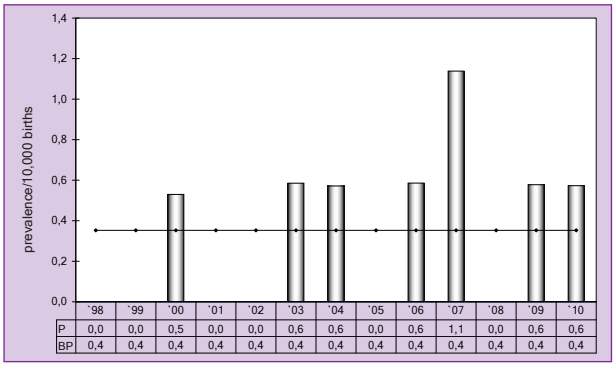


Fig. 29: Development of prevalence/10,000 births with epispadias in the registration area since 1998

In 2010 one epispadias per 17,455 births (8,889 boys) was registered in Saxony-Anhalt.

12.23 Indeterminate Sex (Q56.)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 1 x Dessau-Roßlau	1	2.0	↑
Districts	0	0.0	↓
Saxony-Anhalt	**1**	**0.6**	↔

Indeterminate Sex (1998 to 2009)		
	Basic prevalence /10,000 births	**Confidence interval (CI of 95%) /10,000 births**
Cities	0.42	0.05 - 1.51
Districts	0.86	0.46 - 1.48
Region	0.76	0.42 - 1.25
EUROCAT	0.68	0.14 Sweden* 1.93 SE Ireland**

*/** Centres with lowest resp. highest prevalence/10,000 births

In 2010 one live birth was registered with indeterminate sex in Saxony-Anhalt.
The **prevalence** is at **0.6 per 10,000 births**. This prevalence corresponds to the frequency of appearance registered in other registers such as the EUROCAT centres.

additional Information:

Pregnancy outcome	1 x live birth
Sex	1 x unclear
Number of isolated malformations/MCA	1 x isolated

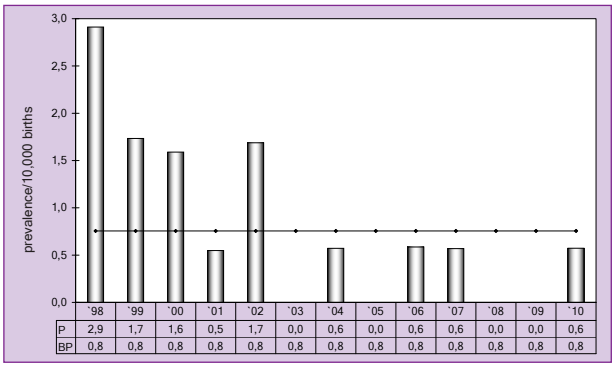


Fig. 30: Development of prevalence/10,000 births with indeterminate sex in the registration area since 1998

In 2010 one child with indeterminate sex per 17,455 births was registered in Saxony-Anhalt.

12.24 Potter Sequence (Q60.6)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 1 x Halle	1	2.0	↔
Districts: 1 x Börde	1	0.8	↓
Saxony-Anhalt	**2**	**1.1**	↓

Potter Sequence (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	1.88	0.86 - 3.56
Districts	2.33	1.62 - 3.23
Region	2.22	1.61 - 2.98
EUROCAT	1.11	0.07 Spain Hospital Network* 6.58 Mainz (Germany)**

*/** Centres with lowest resp. highest prevalence/10,000 births

Two births with potter sequence were registered in 2010. The prevalence of 1.1 per 10,000 births ranges below the expected value and is, compared to the European data of the EUROCAT centres, within the middle range.

One child with a sartan fetopathy registered in 2010 and two cases registered in 2009 were the reason for us to point out the risk of mediacation with ACE inhibitors or AT1-receptor antagonists beyond the first trimenon already in this report and also in our last annual report. Since February 2010 no new case with this typical malformation combination was registered (congenital renal failure, partially combined with a potter sequence and cranial bone hypo-/aplasia).

Both infants showed as main symptom prenatally an oligohydramnios. In one case a renal agenesia bilateral was

additional Information:

Pregnancy outcome	1 x live birth descended within 7 days of life 1 x termination of pregnancy
Sex	1 x male 1 x female
Number of isolated malformations/MCA	2 x MCA

diagnosed intrauterine and the termination of pregnancy took place after 17 weeks of gestation. The diagnosis was confirmed by authopsy.

Malformation combinations (MCA) or superordinated syndromes detected:

- Sartan fetopathy with: hypoplastic cranial bone, dysplastic ears
- Ureter agenesia bilateral, small urinary bladder, micropenis

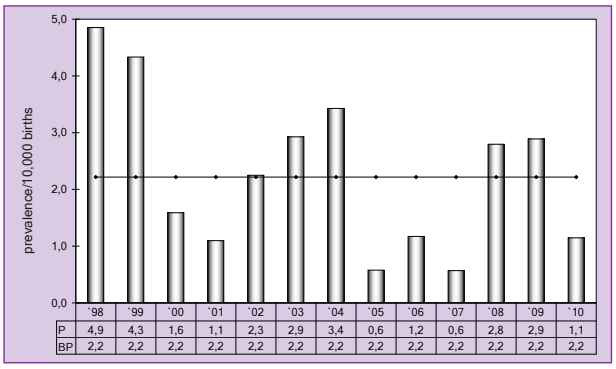


Fig. 31: Development of prevalence/10,000 births with Potter sequence in the registration area since 1998

In 2010 one Potter sequence per 8,728 births was registered in Saxony-Anhalt.

Please note:

What is Sartan fetopathy?

The group of pharmaceuticals sartans were developed from ACE inhibitors. Mainly used in the antyhypertensive therapy, they have a teratogenic effect in case of maternal intake during second and third trimenon of pregnancy.
The suspected pathomechanism is a reduced perfusion of the foetal kidneys resulting in an intrauterine oliguria. Since the amniotic fluid production depends from the second trimenon mainly from the foetal urine production, an oligohydramnios can be diagnosed by prenatal ultrasound screening. This leads into occurrence of a potter sequence with lung and thorax hypoplasia, distorsion of limbs, characteristic facial malformation and other problems. Infants show postnatally an only partial reversible renal failure. Additionally, a hypoplasia/dysplasia of the cranial bone occures in these cases.

12.25 Renal Agenesis, Unilateral (Q60.0/Q60.2)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 1 x Dessau-Roßlau 2 x Halle	3	6.1	↔
Districts: 1 x Altmarkkreis Salzwedel 2 x Harz 1 x Mansfeld-Südharz 1 x Saalekreis 2 x Stendal	7	5.6	↗
Saxony-Anhalt	**10**	**5.7**	↗

Renal Agenesis, Unilateral (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	7.71	5.43 - 10.63
Districts	7.04	5.85 - 8.46
Region	7.20	6.14 - 8.44
EUROCAT	no data available	no data available

Ten births with unilateral renal agenesis were registered in 2010. The **prevalence** of **5.7 per 10,000 births** ranges slightly below the basic prevalence of the years 1998 to 2009.

No EUROCAT data is present for comparison.

In three cases complex malformations, respectively chromosomal disorders were diagnosed prenatally. The terminations of pregnancy took place after 18, 21 and 22 weeks of gestation. The prenatal diagnosis was confirmed by authopsy. In the remaining seven cases the infants were born alive without additional large malformations (with execption of the infant with malformation of ribs).

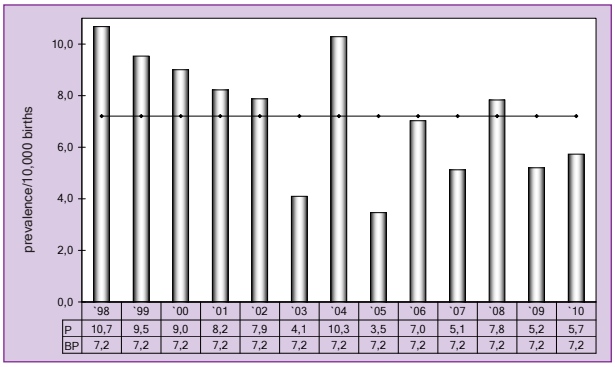
additional Information:

Pregnancy outcome	7 x live births 3 x terminations of pregnancy
Sex	4 x male 6 x female
Number of isolated malformations/MCA	6 x isolated 4 x MCA

In total the renal agenesis ocuured in eight cases on the left side and in two cases on the right side.

Malformation combinations (MCA) or superordinated syndromes detected:

- Trisomy 9 with: microphthalmia, cleft lip with cleft upper jaw and palate left side incl. cleft palate right side, corpus callosum agenesia, VSD, agenesia of arteria pulmonalis, right located aorta ascendens, malrotation and duplex kidney right side, multicystic enlarged ureter right side, hypoplastic pancreas, unlobed lung bilateral, abduction of fingers and adduction of thumbs, malformed feet, large cranial sutures, hypoglossia
- Cyclopia, alobar holoprosencephaly, adrenal gland agenesia left side, hypoplastic lung (right side unlobed, left side indicated fissura horizontalis), malformation of the facial cranial bone, flat thorax, hepatomegaly
- Occipital encephalocele, hypoplasia of aorta, mesenterium ileocolicum commune, hypoplastic gall bladder, cerebellum hypoplasia, micropenis, dilated cerebral ventricle
- Malformation of ribs right side



	`98	`99	`00	`01	`02	`03	`04	`05	`06	`07	`08	`09	`10
P	10,7	9,5	9,0	8,2	7,9	4,1	10,3	3,5	7,0	5,1	7,8	5,2	5,7
BP	7,2	7,2	7,2	7,2	7,2	7,2	7,2	7,2	7,2	7,2	7,2	7,2	7,2

Fig. 32: Development of prevalence/10,000 births with unilateral renal agenesis in the registration area since 1998

In 2010 one renal agenesis, unilateral per 1,746 births was registered in Saxony-Anhalt.

12.26 Cystic Kidney (Q61.1-Q61.9)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 3 x Halle 3 x Magdeburg	6	12.3	↔
Districts: 1 x Altmarkkreis Salzwedel 1 x Anhalt-Bitterfeld 1 x Burgenlandkreis 2 x Börde 1 x Jerichower Land 1 x Mansfeld-Südharz 3 x Salzlandkreis 3 x Wittenberg	13	10.3	↗
Saxony-Anhalt	**19**	**10.9**	↑

Cystic Kidney (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	9.38	6.84 - 12.55
Districts	8.31	7.00 - 9.83
Region	8.57	7.40 - 9.91
EUROCAT	no data available	no data available

*/** Centres with lowest resp. highest prevalence/10,000 births

19 births with cystic kidneys were registered in 2010. All forms of appearance are registered together, excluding the solitary cysts.

The **prevalence** of **10.9 per 10,000 births** ranges above the basic prevalence of the years 1998 to 2009 and above the confidence interval.

In 13 cases a prognistic favourable unilateral mylticystic-dysplastic kidney with partly indicated consecutive hyperplasia of the cystic-free kidney was diagnosed. Within these cases the cystic kidney occurred seven times on the left side and six times on the right side. Additionally, we registered a polycystic bilateral degeneration of kidneys in four cases.

additional Information:

Pregnancy outcome	16 x live births 3 x terminations of pregnancy
Sex	15 x male 3 x female 1 x no information
Number of isolated malformations/MCA	12 x isolated 7 x MCA

At prenatally diagnosed bilateral cystic degeneration of kidneys in combination with additional malformations the termination of pregnancy took place after 18 and 22 weeks of gestation and the fetocide after 31 weeks of gestation.

Malformation combinations (MCA) or superordinated syndromes detected:

- Turner syndrome
- Complex cardiac defect, cleft lip with cleft upper jaw abd palate, craniofacial dysmorphy
- Atrial septal aneurysm with PFO at term infant, hypertrophic kidney left side
- Pes equinovarus congenitus left side, hyperplastic kidney with DUP II.grade left side
- Azygous continuation of the vena cava inferior, hyperplastic kidney left side
- Duplex kidney right side with DUP II.grade
- Ureteropelvic stenosis right side, DUP IV.grade

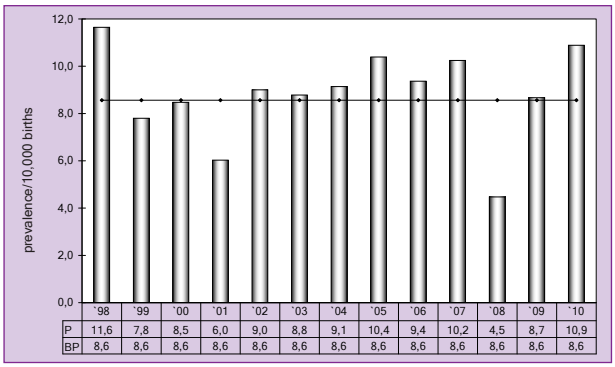


Fig. 33: Development of prevalence/10,000 births with cystic kidneys in the registration area since 1998

In 2010 one cystic kidney per 919 births was diagnosed in Saxony-Anhalt.

12.27 Bladder Exstrophy (Q64.1)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities	0	0.0	↔
Districts	0	0.0	↓
Saxony-Anhalt	**0**	**0.0**	↓

Bladder Exstrophy (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	0.00	0.00 - 0.62
Districts	0.27	0.07 - 0.68
Region	0.20	0.05 - 0.52
EUROCAT	no data available	no data available

No birth with the rarely occuring malformation bladder exstrophy was registered in 2010.
The basic prevalence of the years 1998 to 2009 is at 0.2 per 10,000 births.

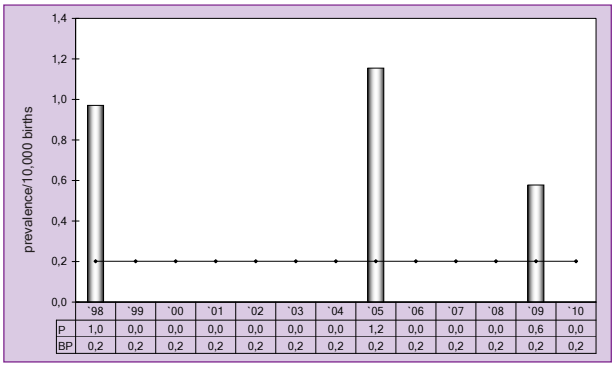


Fig. 34: Development of prevalence/10,000 births with bladder exstropy in the registration area since 1998

In 2010 no birth with a bladder exstrophy was registered in Saxony-Anhalt.

12.28 Preaxial Polydactyly (Q69.1/Q69.2)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 3 x Halle 2 x Magdeburg	5	10.2	↑
Districts: 1 x Altmarkkreis Salzwedel 1 x Burgenlandkreis 1 x Harz 1 x Saalekreis	4	3.2	↘
Saxony-Anhalt	**9**	**5.2**	↔

Preaxial Polydactyly (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	4.38	2.71 - 6.69
Districts	4.25	3.28 - 5.43
Region	4.28	3.42 - 5.29
EUROCAT	no data available	no data available

Nine births with preaxial polydactyly were regsitered in 2010. The prevalence is at 5.2 per 10,000 births and ranges within the upper confidence area.

Comparative EUROCAT data for preaxial polydactyly is not available.

additional Information:

Pregnancy outcome	8 x live births 1 x termination of pregnancy
Sex	2 x male 7 x female
Number of isolated malformations/MCA	7 x isolated 2 x MCA

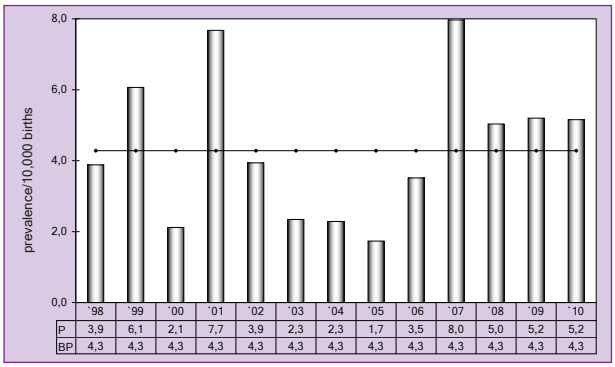
In one case the preaxial polydactyly occurred in combination with an accessory big toe bilateral and equally in four cases with a duplex thumb on the right resp. left side.
In most cases the malformations occured isolated.

In one case a chromosomal disorder was diagnosed and the termination of pregnancy in this case took place after 22 weeks of gestation.

In only one case a positive family history for a preaxial polydactyly is known.

Malformation combinations (MCA) or superordinated syndromes detected:

- Down's syndrome
- Polysyndactyly with: cutaneous syndactyly type III (IV. and V. finger) and osseous syndactyly of toes



	`98	`99	`00	`01	`02	`03	`04	`05	`06	`07	`08	`09	`10
P	3,9	6,1	2,1	7,7	3,9	2,3	2,3	1,7	3,5	8,0	5,0	5,2	5,2
BP	4,3	4,3	4,3	4,3	4,3	4,3	4,3	4,3	4,3	4,3	4,3	4,3	4,3

Fig. 35: Development of prevalence/10,000 births with preaxial polydactyly in the registration area since 1998

In 2010 one preaxial polydactyly per 1,939 births was registered in Saxony-Anhalt.

12.29 Limb Reduction Defects of Upper and Lower Limbs (Q71./Q72./Q73.)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 1 x Halle	1	2.0	↓
Districts: 2 x Altmarkkreis Salzwedel 3 x Burgenlandkreis 1 x Börde 1 x Harz 1 x Stendal	8	6.4	↘
Saxony-Anhalt	**9**	**5.2**	↓

Limb Reduction Defects, in total (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	8.34	5.96 - 11.35
Districts	8.11	6.82 - 9.62
Region	8.16	7.02 - 9.47
EUROCAT	6.03	1.80 SE Ireland* 13.16 Mainz (Germany)**

*/** Centres with lowest resp. highest prevalence/10,000 births

In 2010 nine births with limb reduction defects were regsitered.
The prevalence of 5.2 per 10,000 births falls below the basic prevalence of the years 1998 to 2009 and is outside of the confidence area.

In comparison to the EUROCAT data this value is within the middle range.

additional Information:

Pregnancy outcome	5 x live births 1 x live birth descended within 7 days of life 2 x terminations of pregnancy 1 x stillbirth
Sex	6 x male 3 x female
Number of isolated malformations/MCA	3 x isolated 6 x MCA

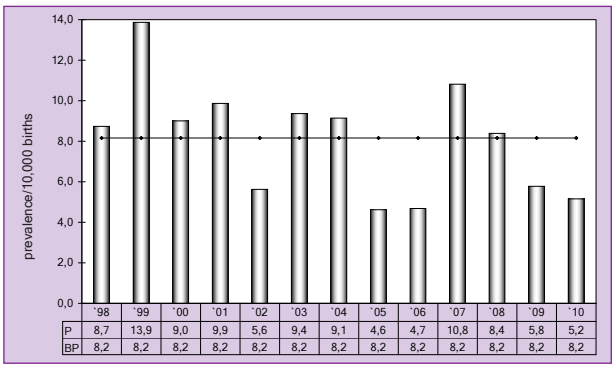
The sex ratio shows an androtropism in 2010.

The most cases occurred with additional malformations. The common VATER association with radial and thumb aplasia occurred in one case. The longitudinal limb malformation of the lower limbs occurred in two cases. In both cases, we were not informed if the malformation was already diagnosed prenatally.

Another two births showed a malformation of the upper limbs, in one case the defect occurred bilateral.
One infant with chromosomal disorder was stillborn after 33 weeks of gestation.

Malformation combinations (MCA) or superordinated syndromes detected:

- Down's syndrome with: single transverse palmar crease and sandal' s gap left side, macroglossia, epicanthus internus
- VATER-association with: hydrocephalus, amniotic strangulation mark basal at the right thumb, VSD, inflected hands and feet, duplex splenic, melded kidney, malformed feet, macrocephaly
- Holoprosencephaly, hypoplasia of the cerebellum, pes equinovarus congenitus left side, broad base of the nose, low-set ears
- Gorlin Goltz syndrome with: omphalocele, microcephaly, cleft of upper lip left side, osseous syndactyly and synphalangy digit 3 and 4 of the right hand, dislocation of lens right side, iris coloboma and coloboma of the papilla bilateral, microcornea left side, duplex kidney bilateral, PFO at term infant, mitral valve insufficiency, retarded hip left side
- Adams-Oliver-Syndrome with: nondescensus testis right side at term infant, truncus arteriosus communis, VSD, aortic valve insufficiency, ASD II, hemangioma
- Stenosis of ureteral orifice and megaureter with DUP IV. grade left side



	`98	`99	`00	`01	`02	`03	`04	`05	`06	`07	`08	`09	`10
P	8,7	13,9	9,0	9,9	5,6	9,4	9,1	4,6	4,7	10,8	8,4	5,8	5,2
BP	8,2	8,2	8,2	8,2	8,2	8,2	8,2	8,2	8,2	8,2	8,2	8,2	8,2

Fig. 36: Development of prevalence/10,000 births with limb reduction defects in total in the registration area since 1998

In 2010 one limb reduction defect per 1,939 births was registered in Saxony-Anhalt.

12.30 Diaphragmatic Hernia (Q79.0/Q79.1)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 2 x Halle	2	4.1	↔
Districts: 1 x Börde 1 x Saalekreis	2	1.6	↔
Saxony-Anhalt	**4**	**2.3**	↔

Diaphragmatic Hernia (1998 to 2009)		
	Basic prevalence /10,000 births	**Confidence interval (CI of 95%) /10,000 births**
Cities	4.59	2.87 - 6.94
Districts	2.26	1.56 - 3.16
Region	2.82	2.13 - 3.66
EUROCAT (Q79.0)	2.33	0.94 Spain Hospital Network* 4.27 Malta**

*/** Centres with lowest resp. highest prevalence/10,000 births

Four births with diaphragmatic hernia were registered in 2010. The **prevalence** of **2.3 per 10,000 births** is below the basic prevalence of the previous years, however, the value is within the confidence area.

In comparison with the EUROCAT data the prevalence is within the middle range.

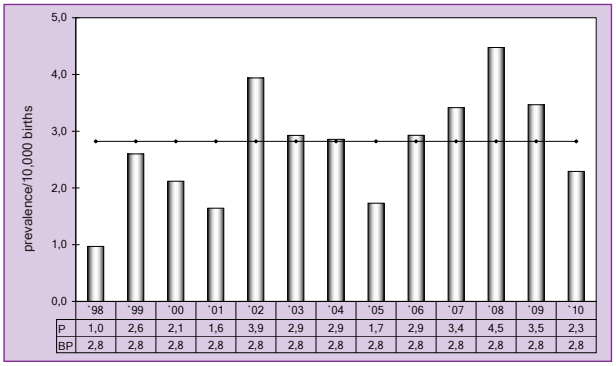
additional Information:

Pregnancy outcome	3 x live births 1 x live birth descended after 7 days of life
Sex	4 x male
Number of isolated malformations/MCA	2 x isolated 2 x MCA

In all four cases the diaphragmatic hernia occurred unilateral. We registered three term infants and one preterm infant, born after 26 weeks of gestation. The infant deceased in the first year of age.

Malformation combinations (MCA) or superordinated syndromes detected:

- PDA at preterm infant (haemodynamical effective), hernia umbilicalis, bilateral hernia inguinalis
- Malformation of aorta



	`98	`99	`00	`01	`02	`03	`04	`05	`06	`07	`08	`09	`10
P	1,0	2,6	2,1	1,6	3,9	2,9	2,9	1,7	2,9	3,4	4,5	3,5	2,3
BP	2,8	2,8	2,8	2,8	2,8	2,8	2,8	2,8	2,8	2,8	2,8	2,8	2,8

Fig. 37: Development of prevalence/10,000 births with diaphragmatic hernia in the registration area since 1998

In 2010 one diaphragmatic Hernia per 4,364 births was registered in Saxony-Anhalt.

12.31 Omphalocele (Q79.2)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 1 x Halle 1 x Magdeburg	2	4.1	↔
Districts: 1 x Burgenlandkreis 1 x Saalekreis 1 x Stendal	3	2.4	↘
Saxony-Anhalt	**5**	**2.9**	↔

Omphalocele (1998 to 2009)		
	Basic prevalence /10,000 births	**Confidence interval (CI of 95%) /10,000 births**
Cities	2.29	1.14 - 4.10
Districts	3.39	2.52 - 4.46
Region	3.12	2.40 - 4.00
EUROCAT	2.45	0.54 Spain Hospital Network* 5.64 Mainz (Germany)**

*/** Centres with lowest resp. highest prevalence/10,000 births

Five births with omphalocele wer registered in 2010. The **prevalence** of **2.9 per 10,000 births** is in 2010 slightly below the basic prevalence and within the confidence area.

In the previous year births with omphalocele were mainly registered in the districts. According to that, seven cases were also registered in 2010 in the districts.

In comparison to the EUROCAT data, the prevalence of Saxony-Anhalt is within the middle range.

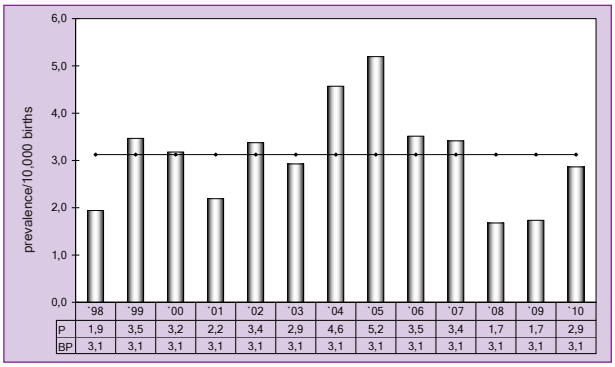
additional Information:

Pregnancy outcome	2 x live births 3 x terminations of pregnancy
Sex	1 x male 3 x female 1 x no information
Number of isolated malformations/MCA	2 x isolated 3 x MCA

Two cases of omphalocele were registered in combination with a trisomy 18, one of these infants was born alive. In one case the omphalocele was diagnosed as part of a Gorlin Goltz syndrome, a complex mesoectodermal malformation syndrome with typical skin atrophy and numerous anomalies of the skeleton, dentition and eyes. This infant was also born alive, the omphalocele and a cleft lip with cleft upper jaw and palate were prenatally diagnosed.

Malformation combinations (MCA) or superordinated syndromes detected:

- Edward's syndrome with: oesophagusatresia with fistula, canalis atrioventricularis communis, persistence of the left vena cava superior, hypoplasia of aorta, horseshoe kidney, overlay of the bowed 3rd and 4th finger by the 2nd and 5th finger bilateral, low-set ears
- Edwards syndrome with: microcephaly, cataract, pes equinovarus congenitus bilateral, osseous syndactyly of toes bilateral, craniofacial dysmorphy, overlay fingers
- Gorlin-Goltz syndrom with: microcephaly, cleft upper lip left side, missing lower leg and foot left side, osseous syndactyly and synphalangy digit 3 and 4 of the right hand, dislocation of lens right side, iris coloboma and coloboma of the papilla bilateral, microcornea left side, duplex kidney bilateral, PFO at term infant, mitral valve insufficiency, retarded hip left side



	`98	`99	`00	`01	`02	`03	`04	`05	`06	`07	`08	`09	`10
P	1,9	3,5	3,2	2,2	3,4	2,9	4,6	5,2	3,5	3,4	1,7	1,7	2,9
BP	3,1	3,1	3,1	3,1	3,1	3,1	3,1	3,1	3,1	3,1	3,1	3,1	3,1

Fig. 38: Development of prevalence/10,000 births with omphalocele in the registration area since 1998

In 2010 one omphalocele per 3,491 births was registered in Saxony-Anhalt.

12.32 Gastroschisis (Q79.3)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 2 x Magdeburg	2	4.1	↔
Districts: 2 x Anhalt-Bitterfeld 2 x Burgenlandkreis 1 x Börde	5	4.0	↔
Saxony-Anhalt	**7**	**4.0**	↔

Gastroschisis (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	4.17	2.55 - 6.44
Districts	3.65	2.75 - 4.76
Region	3.78	2.97 - 4.74
EUROCAT	2.15	0.45 Campania (Italy)* 6.26 Mainz (Germany)**

*/** Centres with lowest resp. highest prevalence/10,000 births

Seven births with gastrochisis were registered in 2010. Therefore the **prevalence** of **4.0 per 10,000 births** is for the first time since 2007 above the basic prevalence of 3.8 per 10,000 births.

The prevalences of Saxony-Anhalt are above the middle range in comparison with the European data. An equal ratio was registered in the previous years, however, the highest prevalence of 8.6 per 10,000 births was registered in 2004.

additional Information:

Pregnancy outcome	6 x live births 1 x termination of pregnancy
Sex	3 x male 3 x female 1 x no information
Number of isolated malformations/MCA	6 x isolated 1 x MCA

In regard to the pregnancy outcome six infants were live births after 26 and 35 weeks of gestation. One termination of pregnancy was carried out after 12 weeks of gestation after the diagnosis of a gastrochisis was confirmed.

Malformation combinations (MCA) or superordinated syndromes detected:

- Microcephalie

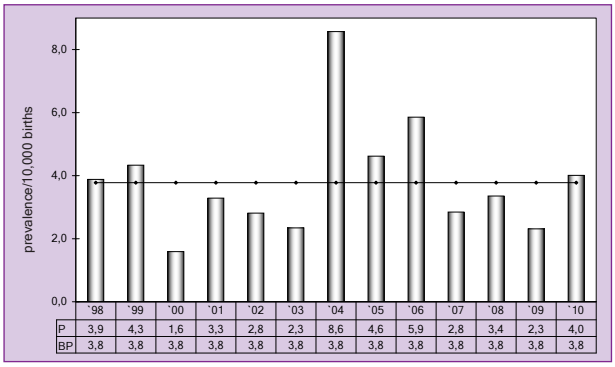


Fig. 39: Development of prevalence/10,000 births with gastroschisis in the registration area since 1998

In 2010 one gastroschisis per 2,494 births was registered in Saxony-Anhalt.

12.33 Prune Belly Sequence (Q79.4)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities	0	0.0	↓
Districts	0	0.0	↓
Saxony-Anhalt	**0**	**0.0**	↓

Prune Belly Sequence (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	0.83	0.23 - 2.13
Districts	0.80	0.41 - 1.39
Region	0.81	0.46 - 1.31
EUROCAT	no data available	no data available

In 2010 no birth with prune belly sequence was registered in Saxony-Anhalt.
As observed in the previous years no births with this rarely appearing malformation are registered in Saxony-Anhalt from time to time. In contrast, in 2002 a high rate of appearance with four registered cases and a prevalence of 3.2 per 10,000 births was observed.

A European comparison is not possible in this case, as no EUROCAT data are present for prune belly sequence.

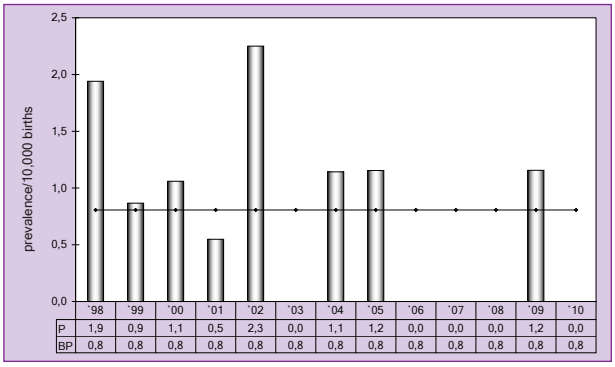


Fig. 40: Development of the prevalence/10,000 births with a prune belly sequence in the registration area since 1998

In 2010 there was no birth with prune belly sequence registered in Saxony-Anhalt.

12.34 Down´s Syndrome - Trisomy 21 (Q90.)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 3 x Dessau-Roßlau 4 x Halle 8 x Magdeburg	15	30.7	↑
Districts: 2 x Altmarkkreis Salzwedel 4 x Anhalt-Bitterfeld 1 x Burgenlandkreis 1 x Börde 6 x Harz 3 x Salzlandkreis 1 x Wittenberg	18	14.3	↔
Saxony-Anhalt	**33**	**18.9**	↗

Down´s Syndrome (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	20.22	16.40 - 24.65
Districts	15.28	13.47 - 17.33
Region	16.48	14.82 - 18.31
EUROCAT	17.89	7.66 S Portugal* 38.54 Paris (France)**

*/** Centres with lowest resp. highest prevalence/10,000 births

As expected, the most frequently appearing chromosomal disorder is the trisomy 21 with 33 registered births in 2010. The prevalence of 18.9 per 10,000 births lies slightly above the prevalence of the previous year and shows a similar high value as in 2008.

A clear difference of the calculated prevalences exists between the districts and major cities. We registered twice as much concerned births in the major cities than in the districts, in relation to the total number of births. Therefore, the prevalence of the major cities is clearly above the confidence area. When regarding the data from the whole Federal State of Saxony-Anhalt, the prevalence is only slightly above the confidence area.

The annual prevalence of 2010 is in comparison with the EUROCAT data within the European middle range. Equally to the previous years the highest prevalence with 38.5 per 10,000 births was registered in Paris.

additional Information:

Pregnancy outcome	10 x live births 2 x live births descended after 7 days of life 20 x terminations of pregnancy 1 x stillbirth
Sex	17 x male 15 x female 1 x no information
Number of isolated malformations/MCA	20 x isolated 13 x MCA

When regarding the pregnancy outcomes, twelve infants with trisomy 21 were born alive. In two of these cases the diagnosis of trisomy 21 was already prenatally confirmed and the families decided to continue the pregnancy. One infant was stillborn after 33 weeks of gestation. In 20 cases of a prenatally confirmed trisomy 21 the termination of pregnancy was carried out between 13 and 21 weeks of gestation.

In 20 cases the trisomy 21 ocurred isolated. 13 births showed additional major malformations as MCA and in eleven cases cardiac defects of different forms were diagnosed.

The sex ratio is nearly balanced. The maternal average age was 35.7 years (27 to 46 years).

Malformation combinations (MCA) or superordinated syndromes detected:

- Anal atresia, VSD, atrophy of the optic nerve bilateral, nondescensus testis at term infant bilateral, ventricular asymmetry, pes adductus bilateral
- Preductal coarctation of aorta, mesenterium ileocolicum commune, malrotation of kidneys
- Microcephaly, unbalanced AV-canal, brachycephaly
- Microcephaly, ASD II, cataract bilateral
- Cor triloculare biatriatum, common atrium, missing lobe of lung right side
- 2 x ASD, VSD, PDA at term infant (1 x with cutaneous syndactylie bilateral)
- 2 x canalis atrioventricularis communis, ASD II (1 x with PDA at preterm infant, plexus cyst bilateral)
- Canalis atrioventricularis communis, triple lobed left lung, underdeveloped nose,
- ASD, retarded hip left side
- Accessoric right thumb
- Hypoplastic left big toe

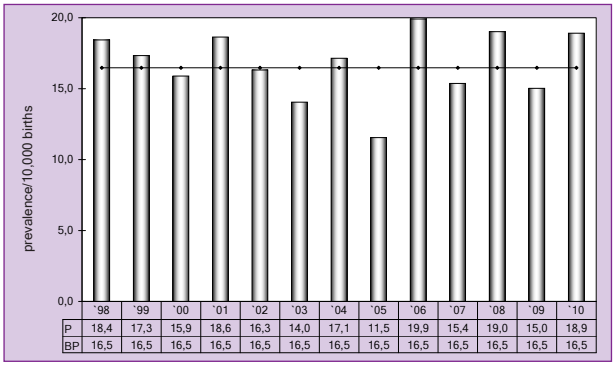


Fig. 41: Development of prevalence/10,000 births with a Down´s syndrome in the registration area since 1998

In 2010 one Down´s Syndrome (Trisomy 21) per 529 births was registered in Saxony-Anhalt.

12.35 Patau Syndrome - Trisomy 13 (Q91.4-Q91.7)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities	0	0.0	↓
Districts: 1 x Harz	1	0.8	↔
Saxony-Anhalt	1	0.6	↘

Patau Syndrome (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	1.67	0.72 - 3.28
Districts	0.93	0.51 - 1.56
Region	1.11	0.69 - 1.68
EUROCAT	1.42	0.26 Zagreb (Croatia)* 3.94 Paris (France)**

*/** Centres with lowest resp. highest prevalence/10,000 births

In 2010 only one birth with trisomy 13 was registered. The prevalence of 0.6 per 10,000 births lies slightly below the confidence area of 1998 to 2009, since the Patau syndrome was not registered in the major cities. The trisomy 13 is the third most common trisomy, however it appears very rarely.

In comparison with the European data, the value is below the European average.

additional Information:

Pregnancy outcome	1 x termination of pregnancy
Sex	1 x male
Number of isolated malformations/MCA	1 x isolated

The patau syndrome was already diagnosed prenatally and the termination of pregnancy was carried our after 15 weeks of gestation.

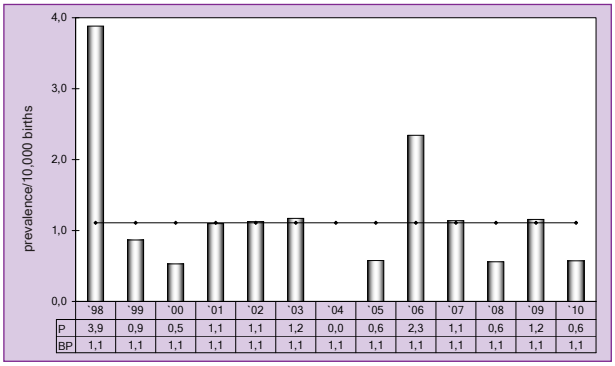


Fig. 42: Development of prevalence/10,000 births with a Patau syndrome in the registration area since 1998

In 2010 one Patau syndrome (Trisomy 13) per 17,455 births was registered in Saxony-Anhalt.

12.36 Edwards-Syndrom - Trisomie 18 (Q91.0-Q91.3)

	Number	Prevalence /10,000 births	Trend in comp. to basic prevalence
Major cities: 3 x Halle 1 x Magdeburg	4	8.2	↑
Districts: 2 x Börde 1 x Saalekreis 1 x Salzlandkreis 2 x Stendal	6	4.8	↗
Saxony-Anhalt	**10**	**5.7**	↑

Edwards-Syndrom (1998 to 2009)		
	Basic prevalence /10,000 births	Confidence interval (CI of 95%) /10,000 births
Cities	3.75	2.22 - 5.93
Districts	3.26	2.41 - 4.30
Region	3.38	2.62 - 4.29
EUROCAT	3.66	0.66 Spain Hospital Network* 12.21 Paris (France)**

*/** Centres with lowest resp. highest prevalence/10,000 births

With ten registered cases of trisomy 18 in 2010 this malformation was registered more frequently in comparison to the previous years. The prevalence of 5.7 per 10,000 births is above the confidence area.

In comparison to the EUROCAT data this value is within the upper third.

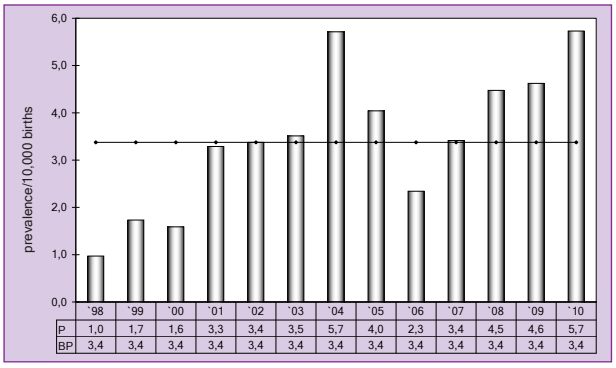
additional Information:

Pregnancy outcome	1 x live birth 9 x terminations of pregnancy
Sex	5 x male 2 x female 3 x no information
Number of isolated malformations/MCA	5 x isolated 5 x MCA

In nine cases of structural anomalies a chromosomal analysis was carried out by prenatal ultrasound screening. In the cases where the diagnosis of trisomy 18 was confirmed prenatally, the termination of pregnancy was carried out between 13 and 19 weeks of gestation.
In five cases we received the authopsy results with additional malformations. In one case one preterm infant of twins was born alive.

Malformation combinations (MCA) or superordinated syndromes detected:

- Holoprosencephaly, microcephaly, cleft upper lip bilateral, VSD, hypoplasia of tentorium
- Omphalocele, oesophageal atresia with fistula, canalis atrioventricularis communis, persistence of the left vena cava superior, hypoplasia of aorta, horseshoe kidney
- Omphalocele, microcephaly, cataract, pes equinovarus congenitus bilateral, osseous syndactyly of toes bilateral
- Coarctation of aorta, cleft lip with cleft upper jaw and palate bilateral, malformed hands, pes equinovarus congenitus left side, dilatation of arteria pulmonalis, malrotation of kidney left side, hypertelorism, pes adductus right side
- Cleft lip with cleft upper jaw and palate right side, pes equinovarus congenitus bilateral



	'98	'99	'00	'01	'02	'03	'04	'05	'06	'07	'08	'09	'10
P	1,0	1,7	1,6	3,3	3,4	3,5	5,7	4,0	2,3	3,4	4,5	4,6	5,7
BP	3,4	3,4	3,4	3,4	3,4	3,4	3,4	3,4	3,4	3,4	3,4	3,4	3,4

Fig. 43: Development of prevalence/10,000 births with Edwards syndrome in the registration area since 1998

In 2010 one Edwards syndrome (Trisomy 18) per 1,746 births was registered in Saxony-Anhalt.

12.37 Indicator Malformations, In Total

In 2010 253 births with one or several indicator malformations were registered.

The Monitoring of Congenital Malformations uses the indicator malformations as exactly defined major malformations according to the definition of the International Clearinghouse for Birth Defects (ICBDSR) (see chapter 12.0). The registered indicator malformations are basis for an international comparison of malformation rates.

In 2010 the total rate of indicator malformations (1.4%) in Saxony-Anhalt was within the middle range of the previous years (related to the total birth population) (2009: 1.3 %; 2008: 1.5%; 2007: 1.4%).

	Number	Percentage (%)
Major cities	91	1.9
Districts	162	1.3
Saxony-Anhalt	**253**	**1.4**

In the following the indicator malformations are regarded individually according to their appearance in the major cities and districts.

The indicator malformation rate is at 1.9% in the major cities of Dessau-Roßlau, Halle und Magdeburg and for the first time since 2005 a clearly higher rate of 1.3% was also registered in the districts.

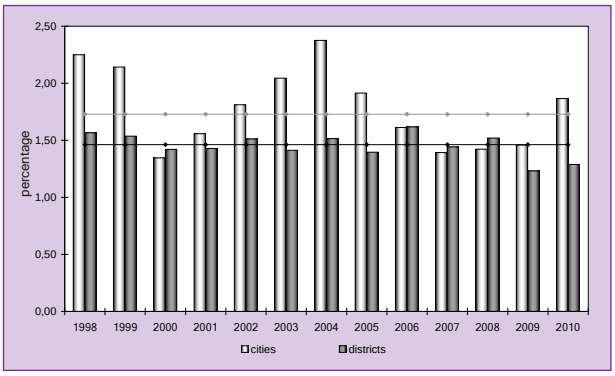


Fig. 44: Total number of indicator malformations of the ICBDSR (1998 to 2010), comparison of frequency (%) in the major cities and districts

The highest incicator malformation rate was registered in Magdeburg with 2.1%, in contrast, a rate of 1.8% was registered in Halle and a rate of 1.5% was registered in Dessau-Roßlau (related to the births in the major cities, see chapter 2 “Births Rate 2010”).

When comparing the single regions, Magdeburg and the district Altmarkreis Salzwedel have a leading position with a rate of 2.1%, followed by Halle and the district Mansfeld-Südharz with a indicator malformation rate of 1.8%.

For the first time a remarkable low malformation rate of 0.8% was registered in the district Saalekreis (in the previous years at least a rate of 1.4% was registered). A possible reason might be the maternal migration for childbirth to Saxony. However, in this context the clearly higher indicator malformation rate of 1.4% which was registered in the bordering district Burgenlandkreis, suprises.

In 2010 a higher appearence of arhinencephaly/holoprosencephaly (4.0 per 10,000 births) was registered in comparison to the basic prevalence of 1.3 per 10,000 births. This can be determined when regarding in total the development of the single indicator malformations prevalences 2010 in contrast to the basic prevalences of the years 1998 to 2009.

The basic prevalence of anorectal atresia shows as well a further increase.

Trisomy 18 attracted attention already in 2004 with a similar high prevalence of 5.7 per 10,000 births in comparison to the basic prevalence of 3.4 per 10,000 births.

The prevalence of transposition of great vessels decreased also in the current year and reaches with 0.6 per 10,000 births a new low-point since 1998.

The same holds true for the hypoplastic left heart syndrome with a new low-point of 0.6 per 10,000 births.

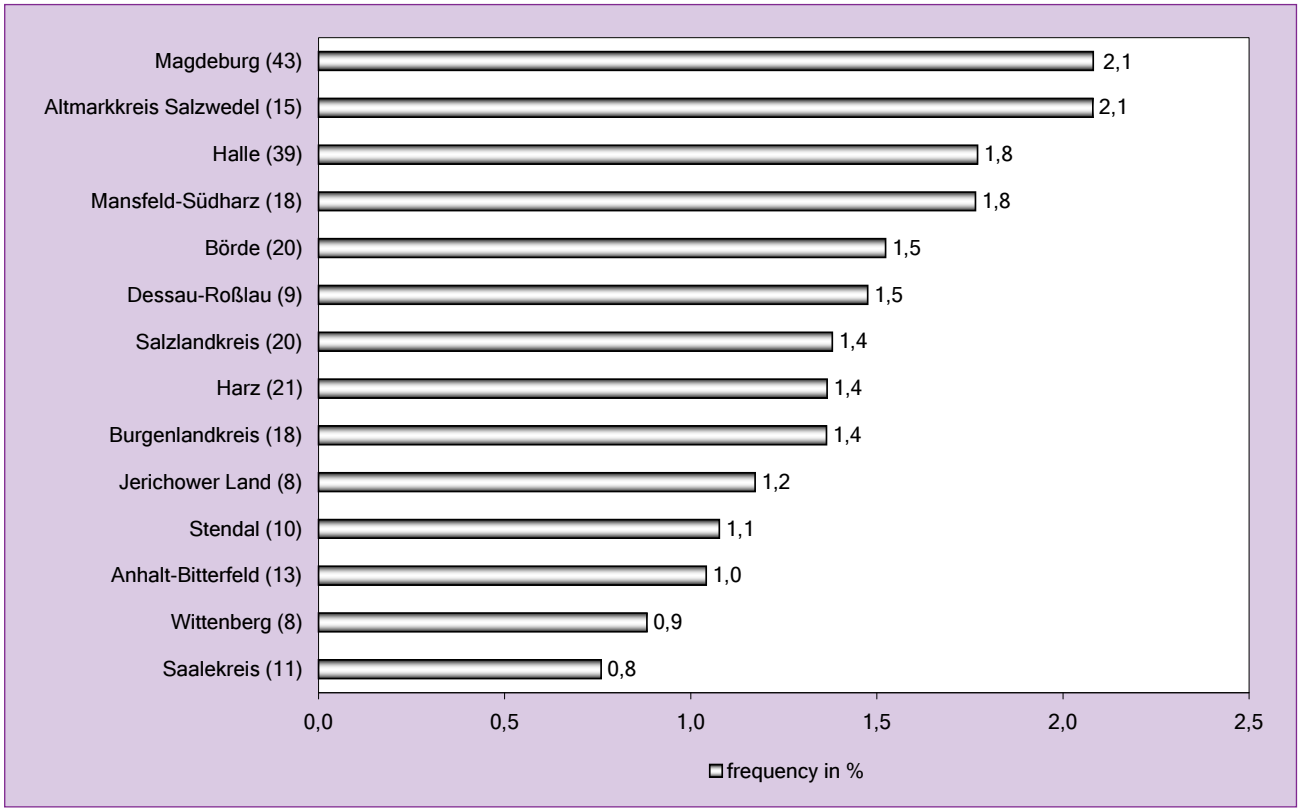


Fig. 45: Total birth rate of indicator malformations in the districts and major cities 2010

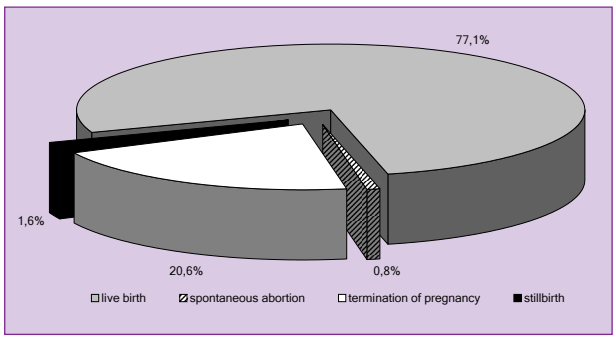


Fig. 46: Pregnancy outcome of births with indicator malformations 2010

In total, 195 of the 253 births with indicator malformations were live births. This corresponds to a percentage of 77.1%. 52 terminations of pregnancy were effected at a present indicator malformation (20.6%) and four infants were stillborn (1.6%). In 0.8% of all cases a spotaneous abortion was registered (2 births).
So we are glad that the rate of live births remained at a high level in comparison to the previous years.

The ratio of live births to terminations of pregnancy remained steady during the last three years.

The rate of stillbirths and spontaneous abortions is still that low, that no analysis of trend is possible.

15 Summary

The 2010 Annual Report of the Federal State of Saxony-Anhalt about the frequency of congenital malformations and anomalies as well as genetically caused diseases contains population-related data with reference to the entire Federal State and is the sixteenth publication of this type. The analysis of data was made in relation to the population according to the offical bith rate provided by the State Statistical Office in Halle.

The total number of live births in Saxony-Anhalt ranged during the last ten years between 18,723 in 2000 and 16,927 in 2006. In 2010 a total number of **17,300 live births** were registered. The number of **63 stillbirths** is very low proportional to the number of live births and confirms the declining trend of stillbirths which was registered during the last 30 years by the Monitoring of Congenital Malformations.

According to the Federal Statistical Office 677,947 infants were live births in Germany in 2010 (2009: 665,126, 2007: 682,514). Only 2.6% of all live births in Germany were born in Saxony-Anhalt.

For the Annual Report 2010 we registered apart from the data of live and still births, data of **67 terminations of pregnancy** and **25 spontaneous abortions after 16 weeks of gestation**. Therefore the prevalence calculations are based on a **total number of 17,455 births** (see chapter 2).

In **580 cases** at least one **major malformation** occured (3.32%). This corresponds to a percentage on the same level as in the previous year (2009: 3.39 %) (see chapter 8).

87.4% of all **births with major malformations** were live births in 2010. This corresponds to the average we registered since 1980 (1980 to 2009: 87.1 %). However, the percentage of live births which died within the first year of life decreased from more than 20% in the beginning of the 1980s to a value below 2% within the last 10 years. A similar development of the numbers of stillbirths can be registered (2010: 0.69 %). The percentage of terminations of pregnancy increased from 1% at the beginning of the 1980s to a value of 9% in the 1990s and is now at 10%. Such a development can be registered due to the improved prenatal diagnosis methods. A percentage of 1.2% of spontaneous abortions was regsitered in 2010 (see chapter 7 and 8).

The analysis of the **frequency of appearance of single diagnoses** shows again that ASD is the most frequent and VSD is the second most frequent single malformation. These two cardiac malformations are followed by dilative uropathy, microcephaly, Down's syndrom and subluxation of hip which occupied from 2000 to 2009 the positions four to seven. The clubfoot (2000 to 2009 position 3) occurred less freuqently in 2010 (position 10) and the PDA occurred more frequently (position 7, 2000 to 2009: position 17) (see chapter 11).

In 2010, 1.45% of all births showed an **indicator malformation** (see chapter 12). This rate is low, however, it is higher than in the previous year (1.29%). Nevertheless, several malformations occurred **more frequently** in comparison to the basic prevalence: microcephaly, arhinencephaly / holoprosencephaly, anophthalmy / microphthalmy, microtia/anotia, coarctation of aorta, choanal atresia, anorectal atresia /-stenosis, hypospadias, cystic kidney, Down's and Edwards syndrome. A **lower prevalence** compared to the basic prevalence was established for the following indicator malformations: spina bifida, hydrocephalus, transposition of great vessels, left heart hypoplasia, cleft palate, small intestine atresia /-stenosis, potter sequence, unilateral renal agenesis, limb reduction defects and patau syndrome. No bladder exstrophy and prune belly sequence were regsitered in 2010 in Saxony-Anhalt.

The Monitoring of Congenital Malformations received data about **64 terminations of pregnancy** in 2010.
Chromosomal aberrations occured more frequently with a value of 59.4% in comparison to the previous years (2009: 47.5 %, 2008: 38.3 %). One case of the very rarely appearing Pallister-Killian syndrome (tetrasomy 12p mosaicism) was registered. The foetus had multiple malformations; however the prenatally determined caryotype was unsuspicious. In 18.7% of all registered malformations defects of the CNS were diagnosed. 84.1% of all terminations of pregnancy were carried out up to 20 weeks of gestation.

27 births suffered from a **genetically caused disease** and in six cases a **sequence, association resp. a complex** was diagnosed in 2010. In seven cases an **embryopathy / fetopathy or congenital infection** occurred. The average maternal average age in the 59 registered cases of a chromosomal aberration was for the first time in more than half of the cases older than 34 years.

The present Annual Report outlines in chapter 16 on the one hand the topic of **30 years of malformation registration**, on the other hand chapter 16.2 deals with the topic of **anorectal malformations**. Within the European comparison we registerd increased prevalences of the anoretal atresia/stenosis with and without fistula. A risk factor for the anorectal malformations might be a high maternal body mass index.

In 2010 the Monitoring of Congenital Malformations received data from 2,313 births from Saxony-Anhalt. At least one major malformation occurred at 580 births. In 230 cases MCA were registered. 285 infants showed only minor malformations. Children without malformations are used as control cases. These data are essential for the analysis of all collected data. When analysing these data, scientifically founded interpretation and calculating of risk factors is possible.

Compilation of the present 2010 Annual Report was only possible due to ongoing voluntary reports about congenital malformations from various medical institutions of Saxony-Anhalt. **We would like to thank all "senders" and hope that this excellent cooperation will continue.**

18 Newborn Hearing Screening 2010

Introduction

After the birth of a child a general newborn hearing screening belongs as from the 01-01-2009 to the recommended early detection examinations. **Aim** of the newborn hearing screening is to **detect congenital hearing disorders at an early stage** (up to the 3rd month of life) and to **initiate the corresponding therapies** (up to the 6th month of life).

Basis for this early detection examination is "Enclosure 6 - early detection examination of hearing disorders at newborns (newborn hearing screening)" of the **Children Directive issued by the Federal Joint Committee (G-BA)**.

The Children Directive determines the **process of the newborn hearing screening** in the following way:

- mesurement of each ear by TEOAE or AABR up to the third day of life (outside of hospital by no later than early detection examination 2 (U2))
- for children at risk AABR obligate
- examinations of preterm infants by no later than calculated term and examinations of ill newborns by no later than third month of life)
- at suspicious first screening, repetition of the examination of both ears by AABR preferably on the same day, but by no later than early detection examination 2 (U2)
- at suspicious finding of the follow-up examination by AABR comprehensive confirmation diagnostics up to the 12th week of life

According to the Children Directive the results of the newborn hearing screening have to be recoded in the "yellow book of examination" of every child. This records are regarded by the responsible paediatrist resp. ENT physician to evaluate if the required diagnosics resp. therapy in case of a hearing disorder was initiated.

The majority of materny clinics in Saxony-Anhalt already offered a newborn hearing screening and tested newborns for hearing disorders before the Children Directive was issued.

The **Monitoring of Congenital Malformations Saxony-Anhalt** cooperates with the Centre for Newborn hearing screening Saxony-Anhalt since 2006 as **tracking center for the newborn hearing screening**.
Step by step we involved the single materny clinics in Saxony-Anhalt and visited the clinics to provid them with information about the hearing screening tracking by use of a tracking-ID and to point out the relevance of this examination.

The **remuneration** of the newborn hearing screening is made within the general births composition payment.
From 01 October 2010 the following scales of charges (GOP) have to be applied for non-hospital examinations and all required follow-up examinations for the settlement of the newborn hearing screening:

01704 surcharge for examination 1 (U1)
surcharge for consultations in the context of the newborn hearing screening according to "Enclosure 6" of the Children Directive of the G-BA; the GOP01711 is applied

01705 Hearing screening
newborn hearing screening according to „Enclosure 6" of the Children Directive of the G-BA

01706 Follow-up examination after suspiciuos first diagnosis
Follow-up AABR according to „Enclosure 6" of the Children Directive of the G-BA after suspiciuos first examination; the GOP 01705 is applied.

Participating Institutions

In 2010 we had 27 materny clinics in Saxony-Anhalt. In all of these clinics a long term newborn hearing screening was and is offered mainly by TEOAE. In 2010 all of these clinics participated in the tracking.

Additionally the screening ID, which is required for the tracking of the hearing screening is used by several midwifes. In this way a newborn hearing screening tracking is also possible for children under care of a midwife (e.g. home childbirths).

In total 26 of the 27 materny clinics participated from 01 January to 31 December 2010 in the hearing screening tracking. The one missing clinic started to participate in the tracking also in the beginning of 2010.

The following table shows the single matery clinics and the periods of time in which the clinics participated in the tracking and shows additionally the number of births with screening-ID.

Materny clinics of Saxony-Anhalt and participation in the hearing screening tracking (ordered by location)

Materny clinics	Trackingperiod 2010	Births in this period*	Births out of trackingperiod**
Klinikum Aschersleben-Staßfurt GmbH	01-01 to 31-12-2010	571	-
Klinikum Bernburg GmbH	01-01 to 31-12-2010	277	-
Gesundheitszentrum Bitterfeld/Wolfen gGmbH	01-01 to 31-12-2010	494	-
MEDIGREIF Kreiskrankenhaus Burg GmbH	01-01 to 31-12-2010	408	-
Städtisches Klinikum Dessau	01-01 to 31-12-2010	813	-
Altmark-Klinikum gGmbH Krankenhaus Gardelegen	01-01 to 31-12-2010	370	-
AMEOS Klinikum St. Salvator Halberstadt	01-01 to 31-12-2010	494	-
Sana Ohre-Klinikum GmbH Haldensleben	01-01 to 31-12-2010	261	-
Krankenhaus St. Elisabeth und St. Barbara Halle	01-01 to 31-12-2010	1,853	-
Universitätsklinikum Halle (Saale)	01-01 to 31-12-2010	1,092	-
Krankenhaus Köthen GmbH	01-01 to 31-12-2010	392	-
Klinik St. Marienstift Magdeburg	01-01 to 31-12-2010	944	-
Klinikum Magdeburg gGmbH	01-01 to 31-12-2010	953	-
Universitätsklinikum Magdeburg A.ö.R.	01-01 to 31-12-2010	1,182	-
Carl-von-Basedow-Klinikum Saalekreis GmbH Merseburg	01-01 to 31-12-2010	682	-
Saale-Unstrut Klinikum Naumburg	01-01 to 31-12-2010	345	-
MEDIGREIF Bördekrankenhaus gGmbH, Neindorf	19-02 to 31-12-2010	178	1
Klinikum Dorothea Christiane Erxleben Quedlinburg gGmbH	01-01 to 31-12-2010	566	-
Altmark-Klinikum gGmbH Krankenhaus Salzwedel	01-01 to 31-12-2010	432	-
Helios Klinik Sangerhausen	01-01 to 31-12-2010	858	-
Klinikum Schönebeck GmbH	01-01 to 31-12-2010	532	-
Johanniter-Krankenhaus Genthin-Stendal gGmbH	01-01 to 31-12-2010	895	-
Asklepios Kliniken Weißenfels-Hohenmölsen GmbH	01-01 to 31-12-2010	506	-
Harz-Klinikum Wernigerode-Blankenburg GmbH	01-01 to 31-12-2010	592	-
Evangelisches Krankenhaus Paul Gerhardt Stift Wittenberg	01-01 to 31-12-2010	573	-
Georgius-Agricola Klinikum Zeitz	01-01 to 31-12-2010	366	-
MEDIGREIF Krankenhaus Anhalt-Zerbst gGmbH	01-01 to 31-12-2010	268	-
		16,897	**1**

Home childbirths and infants born in a birthing center resp. infants not born in Saxony-Anhalt	**157**

* births + multiple births, if no number by the births register was assigned, deducting the stillbirths
** infants which received a screening ID by another clinic or by the tracking centre

In 2010 **16,897 infants** received a scrennning ID after birth in the materny clinic. Therefore these infants are included in the hearing screening tracking.

One infant which was born in the Bördekrankenhaus Neindorf before the clinic started to participate in the tracking, received the sceening ID by the tracking center as in this case a suspicious diagnosis of the hearing screening was reported by a paediatric clinic. This child is not taken into account in the further analysis.

In total, data of **16,897 infants** which were born in Saxony-Anhalt is present for the analysis of the newborn hearing screening. Additionally, data from **157 infants** which were born at home or in a birth center resp. born in other Federal States is present.

17,084 births were registered by the center of newborn screening in Saxony-Anhalt within the newborn screening (see chapter 19). Therefore, a analysis of **99.8%** of this births population in regard to the hearing screening is possible.

Tracking Effort

The tracking of the newborn hearing screening requires an ample organising and personnel effort. The materny clinic has to record the results of the hearing test which are frowarded by mail or fax to the Monitoring of Congenital Malformations. The results are entered here in a special tracking database. In total we received results of **108 senders** in 2010.

The following table shows how much newborns received a screening ID per month and and how many results were forwarded to us per month. It is obvious that at least 1,600 results are reported per month. However in some cases we receive multiple results for one child (e.g. from the materny clinic, paediatric clinic, ENT clinic, ENT physician, paediatrist and from the parents).

Births with screening-ID and number of incoming results

2010	Infants with screening ID	Number of incoming results
January	1.379	1.853
February	1.292	1.667
March	1.354	1.730
April	1.305	1.638
May	1.316	1.835
June	1.514	1.999
July	1.572	2.058
August	1.566	2.062
September	1.584	2.009
October	1.413	1.885
November	1.348	1.806
December	1.411	1.824
total	**17.054**	**22.366**

To carry out the tracking thoroughly, **2,776 letters** were forwarded in 2010 (per infant one up to seven letters). With reference to all infants with screening ID this corresponds to an average of 0.16 letters per infant.

Additionally the parents were contacted by telephone or the parents contacted us by telephone, in total **368 calls** were made (one to four calls per infant).

Results

14,171 infants out of **17,054 infants** with screening ID had an **unsuspicious newborn hearing screening**. In **2,883 cases** the **first hearing test had to be followed-up**, resp. no newborn hearing screening took place in the materny clinic (these cases are regarded as follw-up cases as well). There are numerous reasons why a hearing test did not take place, e.g. ambulant delivery, early discharge from materny clinic, transferation of the child to another clinic, defective hearing screening device.

The **follow-up examination** of the 2,883 infants showed in **2,212 cases** an **unsuspicious result**. The remaining **671 infants** showed again a **suspicious result**. **147** of these 671 infants have a **concluded paediatric audiological confirmation diagnostics**.**187 infants** received **no confirmation diagnostics** and are considered therefore as **lost to follow-up**.

In **259 cases no sceening** was carried out (no reaction of parents to reminder letters or refusal of examination) and in **29 cases** the **status is still pending**, i.e. the examinations were not finished in August 2011 or the tracking process still requires more time. In **13 cases** the **tracking was finished without result** on request of the parents.

In total, the **follow up-examinations** of **158 infants**, born in 2010 could be **finished**. Beside the 147 infants with a suspicious result, 11 infants had an unsuspicious first screening. In **116 cases** a **hearing disorder** could be **excluded**, in **42 cases** a **hearing disorder** was **diagnosed** unilateral/bilateral and the corresponding therapy was initiated.

19 Annual Report 2010 of the Newborn Screening Centre Saxony-Anhalt

according to §14 Note 2 of the valid Children Directive

Head of the Screening centre:
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Introduction

In 2010, again different annual reports had to be filed with the Federal Joint Committee of physicians and health insurances (G-BA) and the German Association for Newborn Screening (number of incoming first-blood tests, incoming of necessary second tests, transmission times, age at point of blood test and others)

20 of 33 suspicious cases for endocrine and metabolic diseases could be confirmed by the corresponding confirmation diagnostics. The care by a metabolic center or an endocrinologist as well as the implemented therapy, when necessary, was controlled and documented.

Our screening laboratory obtained also in 2010 all required certificates for an external quality control of all parameters that are examined during the screening (CDC Atlanta, Deutsche Gesellschaft für Klinische Labordiagnostik). However, our laboratory registered in 2010 a decreased reporting by 10% of only 225 results (2009: 257). In total 27 inpatient institutions, 136 physicians in private practice and 104 midwifes send blood tests of newborns to our screening laboratory in Magdeburg. Probably, the decrease resulted from the commencement of the Gene Diagnistics Act in February 2010.

The annual sender meeting took place on 30 January 2010. Since 68 persons participated in the meeting, we are glad that the meeting has already established as an inherant part of communication between the screening center and our senders. Main topic was the PKU. The form of therapy for this metabolic disorder was outlined by Mrs. Luttat (dietitian at the peadiatric clinic Universitätsklinik Magdeburg), she also outlied the different age groups of the PKU. Dr. Placzek from the metabolic centre of the paediatric clinic Halle presented the following case: PKU with additional cardiologic problems. Dr. Pötzsch, former leader of the Monitoring of Congenital malformations gave an overview about the proceeding in cases of suspected galactosemia. A detailed discussion took place about the presentation of Prof. Dr. Zenker (head of the human genetics clinic at the University Clinic Magdeburg) about the Gene Diagnostics Act. Unfortunately, many questions remained open.

The head of the screening laboratory developed in cooperation with the legal department of the University Clinic a declaration of consent for the newborn screening in accordance with the Gene Diagnostics Act und provided it for our senders. After initial difficulties the cooperation normalised again.

Screening Amplitude

The screening amplitude in Saxony-Anhalt has not changed in comparion to the previous years (PKU, Hypothyroidism, CAH, Galactosemia, biotinidase deficiency, enlarged screening "TMS"). The used methods, reference levels and recall- and detection rates are illustrated in the following two tables:

Tab. 1: Methods/analytics 2010

Parameter	Disease	Method	Reference Value
TSH	Hypothyroidism	Fluorescence immunoassay	<15 mU/l
GALT	Galactosemia	fluorometric	>3.5 U/gHb
BIO	Biotindase deficiency	enzymatic	normal/reduced activity; qualitative method
17OHP	Congenital adrenal hyperplasia (CAH)	Fluorescence immunoassay	depends on gestational age
AC*	see annotation	TMS***	99.9th resp. 0.1th percentile of the normal distribution
AS**	see annotation	TMS***	99.9th resp. 0.1th percentile of the normal distribution

AC* Acylcarnitine - Group of parameter to recognize fattyacid oxidation disorders, organoaciduria and carnitine cycle defects
AS** Amino acids - to recognize aminoacidopathy (PKU, MSUD)
TMS*** Tandem-mass spectrometry

Examination Numbers, Recall Rates and Assured Cases

Table 2 shows the recall rates of the single parameter and the assured cases. 158 recalls had to be done in total in 2010. The two cases of galactosemia are not regarded within this statistics.

Tab. 2: Samples, assured cases, recall-rate 2010

	First test	Second test*	Recall rate** 2010	Assured Cases	Incidence in Saxony-Anhalt 1992-2010
TSH	17.084	695	0,09 %	8	1/3.692
PHE***	17.084	695	0,04 %	7	1/5.571
GALT	17.084	695	0,03 %	0	1/144.961
BIO	17.084	695	0,01 %	0	-
17OHP	17.084	695	0,21 %	2	1/22.072##
AC, AS (TMS)	17.084	695	0,03 %	1 x MCAD#	1/11.869###

* Second transmissions, which were necessary because of an early blood withdrawal at term infant < 36 h or preterm infant < 32 weeks of gestation resp. positive first result (recall)
** Definition of recall: demand of a new blood sample because of a suspicious screenig result, when the first test took place at an age of > 36 h at term infant or >32 weeks of gestation at preterm infant
*** Phe = phenylalanine: parameter for the identification of a phenylketonuria and hyperphenylalaninemia
# MCAD: disorder in metabolizing medium-chain fatty acids
Screening of congenital adrenal hyperplasia syndrome (since 1997 in Saxon-Anhalt)
Enlarged screening (TMS) since May 2001 in Saxony-Anhalt

Registration Rates

The following registration rates were determined in Saxony- Anhalt in 2010: According to the Federal Statistical Office 17,300 children were livebirth in Saxony-Anhalt (according to the residence of the mother).

Tab. 3: Registration rates of first tests

	Number	Difference/Sum
First screening Magdeburg	17.084	
not live in Saxony-Anhalt	726	16.358
Screening refused by parents resp. U2 probably unperceived, no response	8	16.366
Screening in another Federal state*	29	16.395

* only infants were counted whose mother had a residence in Saxony-Anhalt

The discrepancy between the number of live births and screened children amounts to 905.

The data of the Federal Statistical Office are based on the data of the Statistical Office of Saxony-Anhalt. A basis are the registered births by the register offices, these numbers are transmitted by the materny clinics and sorted by residence of the mother. The number of mothers with residence in Saxony-Anhalt, which delivered their children in another Federal State can not be registered in the screening statistics of our center, when the newborn screening took place in another Federal State. When the screening was carried out in another Federal State but the infant was born in Saxony-Anhalt and the mother had her residence in Saxony-Anhalt, this data was included in our statistics when a clear assignment could be ensured.

The control of the second examinations showed the following result: All 659 necessary second examinations (including controls of positive first transmissions) were tracked.

Tab. 4: Registration rates of second tests

	Early withdrawal <36 h.	Preterm infants <32 WOG	Controls of positive first transmissions
Second screening necessary: **659**	379	207	71*
Control in the own laboratory: **626**	359	197	98
Descended before second Screening	2	9	1
Screening in another Federal State	11	1	2
remaining	7	-	1**

* here only real recalls (first transmission >36 h, > 32 weeks of gestation), in two cases no assignment was possible du to missing data
** 1 infant was given a therapy before taking a second blood sample for control

The children, who were screened in another Federal State are living in most cases at the border of other Federal States and the blood withdrawel was taken by midwifes and physicians in private practices, which did not transmit the blood samples to Magdeburg.

The Permanant Committee of the Newborn Hearing Screening of the Working Group for Paediatric Metabolic Disturbances (APS) and for Paediatric Endocrinology (APE) of the German Society of Pediatrics and Adolescent Medicine, the German Society of Neonatology and Paediatric Intensive-Care Medicine (GNPI) and the German Society of the Newborn Screening (DGNS) in collaboration with the German Society of Gynaecology and Obstetrics (DGGG) and the German Society of Perinatal Medicine (DGPM) demand in their Directive of 2002 that an initially required secondary examination should be generally carried out by the originally responsible screening laboratory. Thereby, the process of screening should be easier to control. Unfortunately, this is not possible in every case.

Process Times

Point of Taking Blood Samples

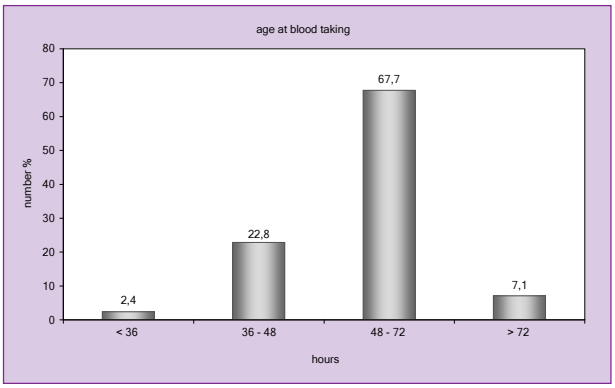

Fig. 1

The optimal point of taking blood samles for the newborn screening (36 -72 hour of life) took place within the required period of time at 92.9% of all cases. At a total number of 7.1%, the taking of blood samples took not place within the required period of time. (2009: 10.5 %).

Note: Only when all the required information was present (date of birth and -time as well as date of blood taking and -time) data of the newborns was registered.

Transmission Time

Figure 2 shows that 37.4% of all transmittals reached the laboratory after more than two days after the blood taking (2009: 37.9 %). 3.4% of all blood samples needed more than four days. (2009: 3.8 %).

Problems with the transmission occured also in 2010, partially because the transmission service changed.

The Children Directive requires a transmission of a pathologic result by the laboratory to the sender by no later than 72 hours after the blood taking. The limiting factor is here the time from the blood taking up to the receipt of the blood sample (delivery time). In this connection we want to point again to Children Directive that requires a transmission of each blood sample at the day of withdrawal.

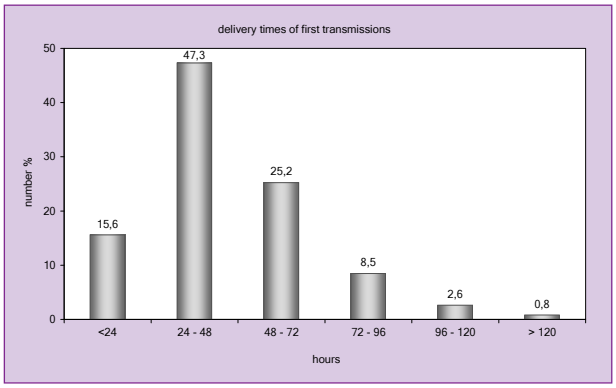

Fig. 2

Transmission of Results

Figure 3 shows the age of the children at the point of result transmission. These data are influenced very much by the time of blood taking, delivery time and time of diagnostics.

At 1.4% (2009: 1,7 %) of all newborns, the screening result was present only after the 8th day of life. In an extreme case, e.g. a typical galactosemia such a result may be too late and the infant might have died. The most blood samples were taken within the required period of time, however they reached the laboratory delayed. In such cases the laboratory calls the attention of the sender by mentioning a corresponding note on the result. Otherwise the situation could not be improved.

The result which is shown by figure 3 regretfully refects also the transmission time of pathologic results (in total 158).

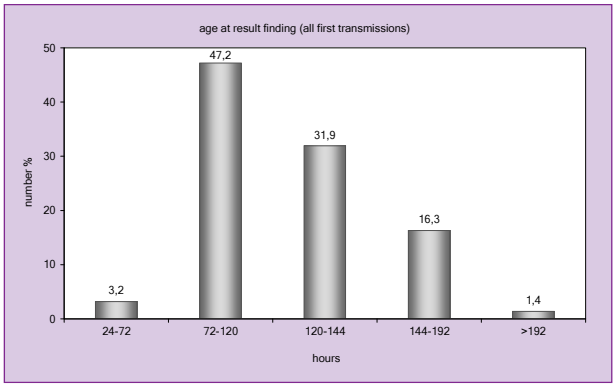

Fig. 3

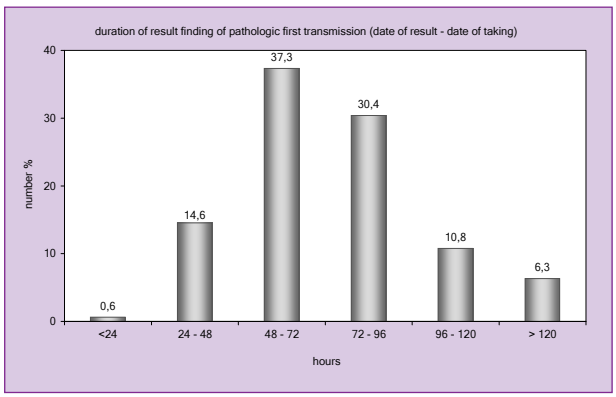

Fig. 4

Figure 4 shows that the 10 findings needed more than 5 days. With reference to the total number of pathological results this can be compared with 2009!

Diagnostics Time of All Results

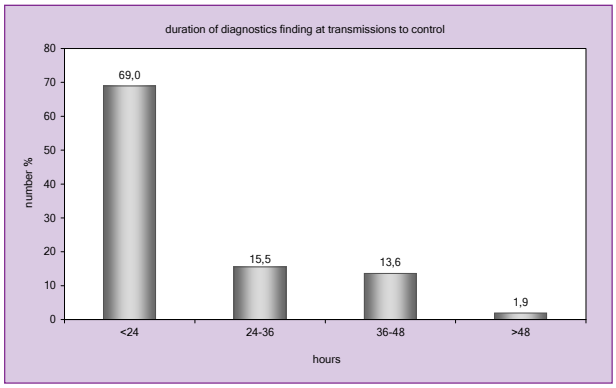

Fig. 5

The diagnostics time is influenced by factors like internal repetitions (necessary, if the first result is classified as pathologic) and malfunction of devices.

98.1% of all results (2009: 97.8%) were finished and transmitted within 48 hours (the printing date counts at normal results and the date of oral transmission at results, which need to be controlled again. The time is documented here at the data set of the child).

Diagnostic Time of Pathological Results

Figure 6 shows the distribution of the diagnostic time of the first transmissions with pathological result. Two results (1.3%) were not present before 72 hours after transmission. The result could not be printed and transmitted earlier due to the missing declaration of consent.

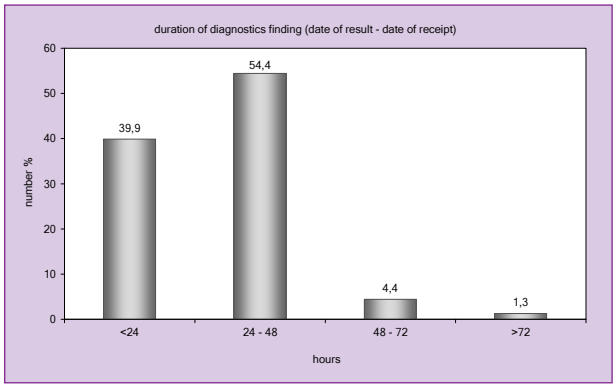

Fig. 6

Figure 7 showes the time period from the oral transmission of the 152 results up to the income of the sample to control. Generally, pathological results are transmitted orally and faxed as partial result after they have been confirmed in the laboratory. All these activities are documented.

In the eleven cases with a response time of more than 120 hours preterm infants were concerned. In these cases, the taking of the sample to control was posponed to a gestational age of 32 weeks (timely second blood taking).

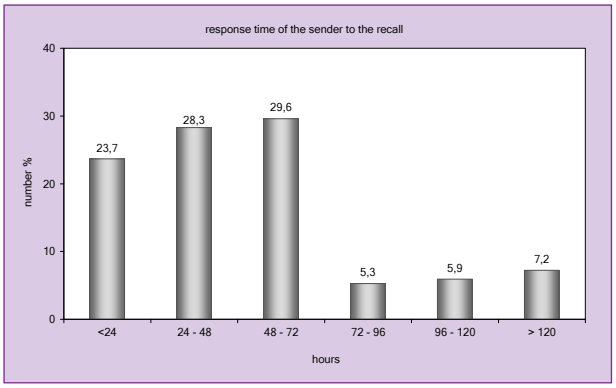

Fig. 7

18 screening suspicious cases were confirmed by confirmation diagnostics. Eight children suffered from a hypothyreosis, seven children suffered from a phenylketonuria /hyperphenylalaninemia (HPA), in two cases a adrenogenital syndrome (AGS) occurred and one infant suffered from from disorders in metabolizing middle-chain fatty acids (MCAD).

Therapy Starting at Patients with Positive Screening

Tab. 5: Diagnosis, confirmation diagnostics and therapie starting

Diagnosis	Confirmation diagnostics	Age at start of therpy
8 x Hypothyroidism	Serum-TSH, T4, sonography	5-8 days
7 x Phenylketonuria	Serum-Phe, BH4-test	7-24 days
2 x CAH	Serum-17-OHP, mutationanalysis, steroidprofile	2-12 days

The children with MCAD do not need a therapy as well as the child suffering from a hyperphenylalaninemia (HPA) does not need a therapy.

Summary

Similar to the previous year, no changes took place in the specifications of the Federal Joint Committee of physicians and health insurances (G-BA). The Children Directive was not revised with regard to the newborn screening until now. After the introduction of the Gene Diagnostics Act, there were initially significant uncertainties and additional work. As it became appearent, every Federal State (screening center) interprets this law in a different way. We informed the senders about our procedure after long discussions with the internal legal department of the University Clinic. The clinics provided us with the information that they do not take blood samples for the newborn screening when no declaration of consent of the parents is present.
Physicians in private practice and midwifes have to send a copy of the declaration to the laboratory and the midwifes have to declare a responsible doctor.

The German Society of the Newborn screening (DGNS) informed the screening centres that the Children Directive will be revised with reference to the Gene Diagnostics Act …deadline open…!

The transmission times (figure 1-7) could be slightly improved again this year.
All patients with a positive first screening result were followed up and the diagnosis assured resp. excluded.
The confirmation of the positive screening result by the attending medical institution and the start of a therapy were also documented in all cases.
An incidence of 1/949 can be calculated for all objective diseases of the newborn screening in Saxony-Anhalt in 2010.

For further information about the metabolic screening centre Magdeburg visit our website

www.stoffwechselzentrum-magdeburg.de

We would like to inform sender, parents and interested people here about the newborn screening and hearing screening and provide downloads. We update our website on a regular basis.