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Ordinance on the model project for comprehensive diagnostics and therapy identification through genome sequencing in cases of rare and oncological diseases

(Genomic Data Ordinance – GenDV)

Genomic Data Ordinance of 8 July 2024 (Federal Law Gazette 2024 I no. 230)

Preamble

The Federal Ministry of Health, after consultation with the Federal Ministry of Education and Research, herewith issues on the basis of section 64e (12) of Book Five of the Social Code (Sozialgesetzbuch V), recast by Article 3 no. 3 letter i of the Act of 22 March 2024 (Federal Law Gazette 2024 I, no. 102), the following:

Section 1
Scope

This Ordinance applies to the model project for comprehensive diagnostics and therapy identification through genome sequencing in cases of rare and oncological diseases pursuant to section 64e of Book Five of the Social Code.

Section 2
Type of data and scope of data to be submitted by healthcare providers

(1) Every healthcare provider that is party to a contract referred to in section 64e (1) sentence 1 of Book Five of the Social Code submits in collated form and for each insured person participating in the model project

1. to the Trust Centre referred to in section 64e (9c) sentence 1 of Book Five of the Social Code so as to identify the healthcare provider responsible for collecting the data, the healthcare provider's name, address and institution code pursuant to section 293 (1) sentence 1 of Book Five of the Social Code,

2. to a genomic data centre referred to in section 64e (10a) sentence 1 of Book Five of the Social Code

   a) so as to identify the healthcare provider responsible for collecting the data, the healthcare provider's name, address and institution code pursuant to section 293 (1) sentence 1 of Book Five of the Social Code as well as the name, telephone number and email address of a contact person assigned by that healthcare provider to the model project,

b) the technical data of the reports referred to in section I of the annex,

c) the genome sequencing data referred to in section II of the annex,

d) the data concerning consent and the participation declaration referred to in section III of the annex and

e) the participant identification number of the healthcare provider referred to in section 4 (1) sentence 2,

3. to a clinical data node referred to in section 64e (10b) sentence 1 of Book Five of the Social Code

a) so as to identify the healthcare provider responsible for collecting the data, the healthcare provider's name, address and institution code pursuant to section 293 (1) sentence 1 of Book Five of the Social Code as well as the name, telephone number and email address of a contact person assigned by that healthcare provider to the model project,

b) the technical data of the reports referred to in section I of the annex,

c) the data concerning consent and the participation declaration referred to in section III of the annex,

d) the general data on the model project referred to in section IV of the annex,

e) the clinical data concerning participation in the model project referred to in section V of the annex,

f) the clinical data concerning previous findings referred to in section VI of the annex,

g) the clinical data for follow-up referred to in section VII of the annex and

h) the participant identification number of the healthcare provider referred to in section 4 (1) sentence 2.

(2) The healthcare provider submits the data referred to in subsection (1) no. 1 to the Trust Centre using the unchangeable part of the insured person's health insurance number referred to in section 290 (1) sentence 2 of Book Five of the Social Code. Pursuant to section 362 (2) sentence 3 of Book Five of the Social Code in conjunction with section 290 (1) sentence 2 of Book Five of the Social Code, the private health insurance companies use for their members the unchangeable part of the insured person's health insurance number as assigned by the Trust Centre referred to in section 290 (2) sentence 2 of Book Five of the Social Code, where available.

Section 3
Specification and updating of data to be submitted

(1) The platform operator regularly compiles recommendations to update and specify the format of the data referred to in the annex and makes these recommendations available to the Federal Ministry of Health by 1 April of each calendar year, starting 1 April 2025. In the recommendations on updating and specifying the format of data relating to oncological diseases referred to in sentence 1, updates to the standardised basic dataset in oncology referred to in section 65c (1) sentence 3 of Book Five of the Social Code are to be observed insofar as these can be applied to the model project.
(2) In the recommendations referred to in subsection (1) sentence 1, the platform operator must observe the state of the art in medical science as well as internationally recognised open standards. The recommendations referred to in subsection (1) sentence 1 are aimed at ensuring that data analyses can be carried out as part of patient care, but also in the context of national, European and international research projects.

(3) When drawing up the recommendations referred to in subsection (1) sentence 1, the platform operator will involve the advisory board referred to in section 64e (9a) sentence 1 of Book Five of the Social Code and, where the format of data from the corresponding fields is concerned,

1. for the field of oncological diseases: the Working Group of German Tumour Centres (*Arbeitsgemeinschaft Deutscher Tumorzentren* – ADT), the Association of Population-based Cancer Registries in Germany (*Gesellschaft der epidemiologischen Krebsregister in Deutschland* – GEKID) and the German Society of Pathology (*Deutsche Gesellschaft für Pathologie*) and

2. for the field of rare diseases: the German Society of Human Genetics (*Deutsche Gesellschaft für Humangenetik* – GfH) and the Working Group of Centers for Rare Diseases in Germany *(Arbeitsgemeinschaft der Zentren für Seltene Erkrankungen*).

Section 4
Data processing by the healthcare providers; data submission deadlines

(1) No later than one month after the healthcare provider becomes a contractual partner to the contract referred to in section 64e (1) sentence 1 of Book Five of the Social Code, each healthcare provider must inform the platform operator of its participation in the model project, providing the name, telephone number and email address of a contact person assigned by that healthcare provider to the model project. As a response to the information referred to in sentence 1, the platform operator issues each healthcare provider with a unique participant identification number. If there are any changes at the healthcare provider that are relevant for participation in the model project, it will inform the platform operator immediately using the participant identification number referred to in sentence 2.
(2) Each healthcare provider generates the working number they need to submit to the Trust Centre pursuant to section 64e (10) sentence 1 no. 1 of Book Five of the Social Code according to a process the healthcare provider determines for itself. The character length of the working number may not exceed 40 characters. It is not permissible for the working number to allow for any inferences to be drawn regarding personal or case-specific details of the insured person participating in the model project. The working number is used exclusively to

1. internally label the dataset to be submitted by the healthcare provider and

2. restore the case reference for data in the cases referred to in section 64e (9c) sentence 7 of Book Five of the Social Code.

(3) The healthcare provider submits the data referred to in section 2 (1) directly upon completing data collection, at the latest three months after completion of data collection. Submission of the data referred to in section 2 (1) is initially carried out

1. for the data referred to in section 2 (1) no. 2: at the latest three months after the operational readiness of a genomic data centre is announced and

2. for the data referred to in section 2 (1) no. 3: at the latest three months after the operational readiness of a clinical data node is announced.

Operational readiness of the sites referred to in sentence 2 is announced by the platform operator on the website of the Federal Institute for Drugs and Medical Devices.

Section 5
Correction procedure

(1) If, during a quality check specified in section 64e (10a) sentence 4 no. 1 and (10b) sentence 4 no. 1 of Book Five of the Social Code, a genomic data centre or clinical data node has doubts regarding the completeness or accuracy of a submitted dataset, the genomic data centre or clinical data node submits a request to the Trust Centre to verify and

complete or correct the dataset together with the genomic dataset pseudonym submitted by the Trust Centre and linked to the dataset pursuant to section 64e (9c) sentence 3 no. 2 of Book Five of the Social Code or together with the clinical dataset pseudonym submitted by the Trust Centre and linked to the dataset pursuant to section 64e (9c) sentence 3 no. 3 of Book Five of the Social Code. The Trust Centre submits the request together with the working number of the dataset to the healthcare provider that submitted the dataset referred to in sentence 1.
(2) To complete or correct a dataset referred to in subsection (1) sentence 1, the healthcare provider that received the request referred to in subsection (1) sentences 1 and 2 submits the data referred to in section 2 (1) no. 1 to the Trust Centre. The Trust Centre generates a transaction number and submits it to the healthcare provider. The healthcare provider submits to the genomic data centre or the clinical data node the completed or corrected data together with the transaction number referred to in sentence 2. The genomic data centre or the clinical data node retrieves the genomic dataset pseudonym or clinical dataset pseudonym referred to in subsection (1) sentence 1 from the Trust Centre using the transaction number referred to in sentence 2, merges it with the completed or corrected data submitted according to sentence 2 and confirms receipt of the completed or corrected data to the healthcare provider.

Section 6
Quality check by genomic data centres and clinical data nodes; reporting confirmation by the platform operator

(1) The platform operator issues a unique participant identification number for every genomic data centre authorised under section 64e (10a) sentence 2 of Book Five of the Social Code and for every clinical data node authorised under section 64e (10b) sentence 2 of Book Five of the Social Code.
(2) After every successfully completed quality check specified in section 64e (10a) sentence 4 no. 1 or subsection (10b) sentence 4 no. 1 of Book Five of the Social Code, the genomic data centres and the clinical data nodes submit to the platform operator without delay an assessment report as well as the transaction number submitted by the healthcare provider as specified in section 64e subsection (10) sentence 1 no. 2 of Book Five of the Social Code (genomic transaction number) or the transaction number submitted by the healthcare provider as specified in section 64e (10) sentence 1 no. 3 of Book Five of the Social Code (clinical transaction number). The assessment report contains

1. when submitted via a genomic data centre

   a) the date of receiving the data specified in section 2 (1) no. 2,

   b) the type of report referred to in section (I) no. 2 of the annex,

   c) the information that the respective data were submitted fully,

   d) information as to whether whole genome sequencing, exome sequencing, sequencing of large gene panels that relate to a specific disease or no sequencing has taken place and

   e) the participant identification number of the healthcare provider referred to in section 4 (1) sentence 2,

2. when submitted via a clinical data node

   a) the date of receiving the data referred to in section 2 (1) no. 3 letters a to f and h,

   b) the type of report referred to in section (I) no. 2 of the annex,

   c) the information that the respective data were submitted fully and

d) the participant identification number of the healthcare provider referred to in section 4 (1) sentence 2.

(3) In response to the submission specified in subsection (2) sentence 1, the platform operator immediately generates reporting confirmations and submits these together with the genomic transaction number or the clinical transaction number to the relevant healthcare provider.
(4) The reporting confirmations referred to in subsection (3) contain

1. an identification number to be assigned by the platform operator for the reporting confirmation,
2. the date of the reporting confirmation,
3. the type of report referred to in section (I) no. 2 of the annex,
4. the information that the respective data were fully submitted,
5. information as to whether whole genome sequencing, exome sequencing, sequencing of large gene panels that relate to a specific disease or no sequencing has taken place,
6. information as to whether clinical or genomic data have been collected and
7. information as whether the particular case concerns an oncological or rare disease.

(5) The platform operator must delete the genomic transaction number and the clinical transaction number after submission to a healthcare provider.

Section 7
Pseudonymisation process; exercise of duties by the Trust Centre

(1) The Robert Koch Institute carries out the duties of the Trust Centre independently and separately from its other tasks. The Trust Centre is separated spatially, organisationally, technically and in terms of personnel from the platform operator, the genomic data centres and the clinical data nodes.
(2) The data submissions specified in sections 2 and 5 take place via a secure mode of submission. The requirements for secure submission of the data specified in section 2 (1) no. 1, subsection (2), section 5 (1) sentences 1 and 2 are determined by the Trust Centre according to the state of the art and published up to date on the website of the Robert Koch Institute.
(3) In response to the submission of the data specified in section 2 (1) no. 1, the Trust Centre submits to the healthcare provider that submitted the data specified in section 2 (1) no. 1 f

1. genomic transaction number and
2. clinical transaction number.

(4) It is not permissible for the genomic transaction number and the clinical transaction number to allow for inferences to be drawn regarding any personal or case-specific details of an insured person participating in the model project.
(5) The Trust Centre determines using a self-selected process whether the submission of a genomic dataset pseudonym to a genomic data centre and a clinical dataset pseudonym to a clinical data node was performed without errors.

Section 8
Entry into force

This Ordinance enters into force on the day following its promulgation.

Closing remarks

The Bundesrat has given its approval.

Annex
Type of data and scope of data

(Publication reference: Federal Law Gazette 2024, no. 230, pp. 6-10)

I. Technical data of the reports to be submitted to a genomic data centre and a clinical data node)

1. Date of report
2. Type of reports
   a) Initial reports
   b) Follow-up reports
   c) Additional reports
   d) Corrections

II. Genome sequencing data (to be submitted to a genomic data centre, for all healthcare providers)

1. Raw data equivalents
   a) Whole genome sequencing
   b) Exome sequencing
   c) Sequencing of large gene panels that relate to a specific disease
2. Additional sequencing data
   a) Manufacturer and model of sequencer
   b) Manufacturer and designation of flow cell and sequencing kit
   c) Run information (sequencing depth, barcode definitions, read length and read direction (single-read or paired-end sequencing)), manufacturer and designation of sequencing library preparation kit
   d) Date of sample
   e) Sample type (e.g. tumour-derived deoxyribonucleic acid (DNA) and/or germ line), source material (e.g. blood, tissue) and preservation method (e.g. fresh or fresh frozen or Formalin-Fixed Paraffin-Embedded (FFPE))
   f) Target region in case of enrichment of genomic regions (information on target region and kit/manufacturer of the enrichment method), reference genome (e.g. hg19); in the event of tumour sequencing, also the fraction of tumour cells within the sample
3. Data on the evaluation methodology (e.g. variants found in the coding DNA after performing whole genome sequencing)

III. Data relating to consent and the participation declaration (to be submitted to a genomic data centre and a clinical data node)

1. Participation in model project
   a) Insured person (patient identification number or pseudonym)

b) Healthcare provider (receiving facility)

c) Date of participation declaration

d) Version of participation declaration

e) Scope of participation declaration

aa) Participation declaration for model project and consent to genome sequencing

bb) Consent to case identification to facilitate professional exchange between clinicians

cc) Consent to re-identifying of the insured person's data and being contacted if new research findings emerge

f) Date of withdrawal of participation declaration

g) Scope of withdrawal of participation declaration

aa) Participation declaration for model project and consent to genome sequencing

bb) Consent to case identification to facilitate professional exchange between clinicians

cc) Consent to re-identifying of the insured person's data and being contacted if new research findings emerge

2. Research participation consent

a) Date of withdrawal of consent

b) Scope of withdrawal of consent

IV. General data on the model project (to be submitted to a clinical data node)

1. Gender

2. Month and year of birth

3. The first five digits of the official municipality code of the patient's domicile at the time of the initial enrolment in the model project

4. Presentation of the insured person to participate in the model project

a) Decision by multidisciplinary case conference in the area of diagnostics to admit the insured person to the model project

b) Date of multidisciplinary case conference

V. Clinical data concerning participation in the model project (to be submitted to a clinical data node)

1. For oncological diseases

a) Diagnosis

aa) Tumour diagnosis according to International Statistical Classification of Diseases and Related Health Problems (ICD)

bb) Date of initial diagnosis (in relation to previously mentioned diagnosis)

cc) Eastern Cooperative Oncology Group (ECOG) performance status

dd) Additionally for hereditary cancer syndromes: Phenotyping based on Human Phenotype Ontology (HPO)

b) Pathological finding

aa) Histological finding ICD-O-3

bb) Differentiation grade

c) Tumour stage

aa) According to the current classification of malign tumours by tumour, node and metastases (TNM) staging system to depict the tumour's size and degree of metastasis including the code as determined by the TNM staging system

bb) According to the diagnosis-specific classification of tumour types, where the TNM staging system does not apply

d) Decision by the multidisciplinary case conference

aa) Date of case conference

bb) Recommendation for participation in a clinical study

cc) Recommendation for systemic treatment (outside of a clinical study)

aaa) Recommendation for On-Label Use

bbb) Recommendation for Off-Label Use

ccc) Recommendation for a compassionate use programme

dd) Recommendation for human genetics counselling

ee) Recommendation for diagnostic re-evaluation

ff) Type of recommended treatment (including the preventive measures against hereditary cancer syndromes)

gg) Active ingredient

hh) Level of Evidence

ii) Supporting molecular alteration according to standard (e.g. Online Mendelian Inheritance in Man (OMIM) or HUGO Gene Nomenclature Committee (HGNC))

aaa) Within the coding DNA

bbb) In regulatory regions

ccc) Outside the coding DNA and regulatory regions

jj) Recommendation for surgery

2. For rare diseases

a) Phenotyping

aa) Phenotyping based on the HPO

bb) Month and year first HPO-coded symptom appeared

b) Decision by the multidisciplinary case conference

aa) Date of case conference

bb) Genomic diagnostics upon which the recommendation is based

aaa) Singleton genome

bbb) Duo genome

ccc) Trio genome

cc) Assessment of genetic diagnostics

aaa) No genetic diagnosis

bbb) Tentative genetic diagnosis

ccc) Further genetic diagnostic tests recommended

ddd) Genetic diagnosis secured

eee) Clinical phenotype only partially resolved

dd) Diagnosis

aaa) According to the International Classification of Diseases in the respective version issued by the Federal Institute for Drugs and Medical Devices on behalf of the Federal Ministry of Health and enacted by the Federal Ministry of Health

bbb) According to the classification of rare diseases (ORPHAcode)

ccc) Additional international classifications after promulgation by the Federal Institute for Drugs and Medical Devices

ddd) Details about possible health impairments according to the Gross Motor Function Classification System (GMFCS)

ee) Recommendation for participation in a clinical study

ff) Recommendation for human genetics counselling

gg) Recommendation for diagnostic re-evaluation

hh) Recommendation of a treatment, type of treatment, underlying genetic variant(s)

ii) supporting molecular alteration according to standard (e.g. OMIM or HGNC)

aaa) Within the coding DNA

bbb) In regulatory regions

ccc) Outside the coding DNA and regulatory regions

jj) Additional recommendation for clinical management

VI. Clinical data on previous findings (to be submitted to a clinical data node)

1. For oncological diseases

a) Case-related, previous genetic findings

aa) Type of diagnostic test

aaa) Whole genome sequencing

bbb) Exome sequencing

ccc) Sequencing of large gene panels that relate to a specific disease

ddd) Sequencing of small gene panels that relate to a specific disease

eee) Single gene testing

bb) Date of diagnostic test

cc) Result of diagnostic test

b) Additionally for non-hereditary cancer syndromes: case-related previous treatment

aa) Type of treatment

bb) Active ingredient

cc) Start date

dd) End date

ee) Date of progress

ff) Response to therapy (best response)

aaa) Progression

bbb) Stable disease

ccc) Partial remission

ddd) Full remission

2. For rare diseases

a) Case-related, previous genetic findings

aa) Type of diagnostic test

aaa) Array

bbb) Single gene tests

ccc) Sequencing of small gene panels that relate to a specific disease

ddd) Chromosome analysis

eee) Sequencing of large gene panels that relate to a specific disease

fff) Exome analysis

ggg) Others

bb) Date of diagnostic test

cc) Result of diagnostic test

b) Case-related previous treatment

aa) Number of inpatient treatment episodes in the last five years and estimated duration in days

bb) Month and year of first contact with a specialised centre for rare diseases

VII. Clinical data on follow-up reporting (to be submitted to a clinical data node)

1. For oncological diseases

a) Implementation of a treatment recommendation by the case conference (including the preventive measures in case of hereditary cancer syndromes)

b) Treatment

aa) Start date of treatment

bb) End date of treatment

cc) Type of treatment

dd) Active ingredient

c) Response to therapy (best response)

aa) Recording date

bb) Data source of response to therapy

cc) Progression

dd) Stable disease

ee) Partial remission

ff) Full remission

gg) Additionally for hereditary cancer syndromes: Emergence of metachronous tumour diseases with diagnosis code

d) ECOG performance status

e) General status regarding clinical follow-up

aa) Vital status

bb) Date of last contact

cc) Day, month and year of death

2. For rare diseases

a) Documentation of follow-up visit

aa) Date

bb) Details on changes in the phenotype based on the HPO

cc) Details on changes in health impairments according to the GMFCS

dd) Diagnosis performed

ee) Supporting data on the course of the illness

b) General status regarding clinical follow-up

aa) Vital status

bb) Date of last contact

cc) Month and year of death