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**State of the Art of Rare Disease
Activities in EU Member States and Other
European Countries:
Question Survey and Data Collection
Methods**

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2. Question Mapping

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The Report on the State of the Art of Rare Disease (RD) activities in Europe is a well-established resource providing valuable, detailed information for all stakeholders in the field of RD and orphan medicinal products. It highlights activities and progress at both the European Union (EU) and Member State (MS) levels and provides insight as to what a country has achieved over the course of each year. Under the EUCERD Joint Action the report was produced annually, by the INSERM team in Paris. This was a substantial document, downloaded 15,000 times per annum and divided into five volumes. Under the new Joint Action, RD-ACTION, production of the State of the Art has moved to Newcastle University. The report will become a more streamlined, online resource which will allow the information to be accessed more easily and updated more regularly (although an overview report will still be produced once a year).

In order to collect and collate this information RD-ACTION has been researching electronic methods of data collection and trying to find ways of streamlining the process and gathering data more efficiently – it was agreed that respondents should complete an online questionnaire. Using the *Guideline document for elaborating information on the State of the Art of Rare Disease activities in your country* (the ‘prompt’ document used 2012-2014) and the *EUCERD Recommendations on Core Indicators for Rare Disease National Plans/Strategies*, a new list of questions has been compiled. These questions include those defined in the Core indicators Recommendations table; however, they have been supplemented to enable additional pertinent information to be obtained. Questions take different forms (i.e. tick boxes or free text comments boxes) and respondents are provided with a means of elaborating further if they wish to do so.

Data will be provided by small Data Contributing Committees (DCCs), composed of the representative of the Commission Expert Group on Rare Diseases, a representative of the National Alliance of patient organisations, and a representative of the Orphanet team. We have mapped each question to illustrate its structure within the online survey. Below is a key which can provide further information on the colour coding of the maps.

Colour/Shape	Meaning
Orange	Question
Blue	Answer – detailing the form of answer required
Green	Proceed to next section of the survey
Yellow arrow	Direction to take through survey

The mapped questions in this document illustrate **all possible questions** within the matrix of the questionnaire; however, different responses will initiate different pathways, streamlining the process of completing the survey.

The medium for completing will be Lime Survey, for the following reasons:

- an electronic means of capturing this information is required, as there is no budget to enable the DCCs to meet physically
- sharing Word documents and updating these between stakeholders is often difficult and time-consuming
- some institutions (especially Competent National Authorities) do not permit access to Google Drive and similar collaborative tools
- free online tools such as Survey Monkey do not have the functionality to allow several users to contribute to the same form, whereas Lime Survey enables this (and also provides translation services).

The DCCs will be given a summary of the information provided for the 2013 report which will allow them to reflect on the changes from Dec 2013 to the present day in their country, answer the questions more quickly and ensure the information they provide for the new report is up-to-date and highlights their achievements.

Question:

Does your country's NP/NS for rare diseases address the coding of RDs in health information systems?

Answer:

Free Text Comments Box

Question: (If answered YES to the financial support question)

Please stipulate the amount of public funds allocated for the NP/NS per year (Euros):

Answer: (Free text box to enter number)

Amount

Answer: (Free text box to enter number)

Amount per million inhabitants – enter number

Answer: (Free text box to enter number)

Value partially available

Answer: (Free text box to enter number)

Funding for the NP/NS activities are incorporated into the general budget – enter number

Question: (If answered NO to financial support question)

Question:

Is there a dedicated body (expert advisory group) to oversee drafting or implementing of the NP/NS, or to evaluate the impact of the NP/NS?

Answer: (Tick Box)

i. Yes this exists, is multi-stakeholder (including patients) and meets regularly.

Answer: (Tick Box)

ii. Yes this exists, is multi-stakeholder (including patients) and is functioning (irregular meetings and activities).

Answer: (Tick Box)

iii. Yes this exists but is partially functioning and does not include all relevant stakeholders.

Answer: (Tick Box)

iv. No, there is no dedicated advisory body.

Question: (If selecting i, ii, or iii):

Please provide details of this body (including its membership, level of patient participation, and methods of implementing and evaluating the NP/NS)

Answer:

Free Text Comments Box

Please proceed to next section

Question:

Please highlight the major successes of the NP/NS to date (e.g. prominent measures implemented under the NP/NS).

Answer:

Free Text Comments Box

Please proceed to next section

B. Rare Disease Registration

Question:

Does your country have a national or regional registry for rare diseases?

Answer: (Tick Box)

a) National RD registry in place

Answer: (Tick Box)

b) Regional RD registry(ies) in place

Answer: (Tick Box)

c) Both national and regional registries for RD

Answer: (Tick Box)

d) Neither national nor regional RD registries exist, but disease specific registries are available.

Answer: (Tick Box)

e) No registries exist for RD in my country.

Question: (If answered a, b or c)

Please provide links to any laws or regulations governing RD registries (of any kind) in your country.

Answer:

Free Text Comments Box

Question:

Please summarise the status quo of RD registration in your country i.e. summarise the history and operations of the national/regional registry (please also summarise the status quo of relevant disease-specific registries) (limit 500 words)

Answer:

Free Text Comments Box

Question:

How are the registries funded?

Answer:

Free Text Comments Box

Question:

If possible, please indicate the (approximate) number of patients currently registered.

Answer:

Free Text Comments Box

Question:

Do your registries code RD using OrphaCode (if not, please describe the coding systems used).

Question: (If answered d)

Are there any laws/regulations in place governing RD registration?

Answer:

Free Text Comments Box

Question:

Please provide a summary of disease-specific national registries (e.g. which diseases/groups of diseases are addressed, whether these are clinician-led, patient-led, etc.)

Answer:

Free Text Comments Box

Please proceed to next section

Question: (If answered e)

Please summarise the status quo re. Registries for RD in your country, including any plans to establish a national registry in the future.

Answer:

Free Text Comments Box

Please proceed to next section

Answer:

Free Text Comments Box

Question:

Regarding your national/regional level registries only: Please provide brief details.

Do you have any future plans/capacity to integrate data?

How are data from the national/regional registries used?

Answer:

Free Text Comments Box

Please proceed to next section

C. Definition of a Rare Disease

Question:

Has your country adopted the European Commission definition of a rare disease?*

**The EU defines 'rare disease' as those with a prevalence of no more than 5 patients per 10,000 persons. This definition is laid down in Regulation EC no. 141/2000 on Orphan Medicinal Products, Directive 2011/24/EU on Cross Border Healthcare as well as in the Council Recommendation on an action in the field of rare diseases of 8 June 2009.*

Answer. (Tick Box)

a) Yes, and the NP/NS (if existing) espouses this definition.

Answer. (Tick Box)

b) Yes, but the NP/NS measures use a different definition (please specify in free text comments box)

Answer. (Tick Box)

c) No

Answer.

Free text comments box

Question: (if answered C)

Please explain how rare diseases are defined in your country

Question:

Are there instances where a different definition is used? (e.g. for reimbursement purposes)

Answer.

Free text comments box

Answer.

Free text comments box

Please proceed to next section

Please proceed to next section

D. Neonatal Screening

Question:

Please summarise the policies and legislation pertaining to new born/neonatal screening in your country.

Answer:

Free Text Comments Box

Question:

How many rare diseases are covered in the neonatal screening programme in your country? (Please list)

Answer:

Free text comments box

Please proceed to next section

E. Genetic Testing

Question:

Please summarise the organisation of genetic testing in your country. To include:

- Links to any relevant national laws/regulations governing the use of genetic testing.*
- Is there a summary/list/registry of reference laboratories providing specific genetic tests?*
- Do national laboratories provide details to the Orphanet database?*
- Are there specific provisions in place for the reimbursement of tests?*

Answer:

Free Text Comments Box

Question:

*Please describe any policies in place to facilitate **Cross Border** genetic testing for Rare Diseases.*

Answer:

Free Text Comments Box

Please proceed to next section

F. Centres of Expertise

Question:

Is there an official policy for designating Centres of Expertise for RD in your country?

Answer: (Tick Box)

Yes

Answer: (Tick Box)

No

Question:

Is there a National Policy, a Regional Policy or both?

Answer: (Tick Box)

A) National policy

Answer: (Tick Box)

B) Regional policy

Answer: (Tick Box)

C) My country has adopted a national and regional designation process

Question: If A) or C) selected

i. Is this formal National Policy for designating centres of expertise for RD fully implemented (i.e. centres have been officially designated) or partially implemented?

Answer:

Free Text Comments Box

Question: If A) or C) selected

ii. Please describe the national criteria used to designate a CE for RD (e.g. are these in accordance with the EUCERD criteria?)

Answer:

Free Text Comments Box

Question:

Please state the number of Centres of Expertise complying with the National or Regional Policy

Answer:

Free text to specify number

Question:

Please describe how RD expertise is identified at present.

Are there any plans to adopt a formal National Policy for establishing Centres of Expertise for rare disease?

Answer:

Free Text Comments Box

Question:

Are there any plans to adopt a formal National Policy for establishing Centres of Expertise for rare disease?

Answer:

Free Text Comments Box

Please proceed to next section

Question:

Please state the number of CEs divided by million inhabitants

Answer:

Free text to specify number

Question:

Please state the number of CEs fulfilling the EUCERD criteria

Answer:

Free text to specify number

Question:

Do CEs ensure a holistic approach to care (e.g. do they liaise with social services?)

Answer:

Free text reply

Please proceed to next section

G. European Reference Networks (ERNs)

Question:

Does your country have a formal process in place for endorsing Health Care Providers (CEs) to participate as members or coordinators of an ERN?

Answer: (tick box)

Yes

Answer: (tick box)

No

Question:

*Are any National Health Care Providers (HCPs) in your country currently **coordinating** an ERN?*

Answer: (tick box)

Yes

Answer: (tick box)

No

Answer: (tick box)

Don't know

Answer: Free text comments box for name of the ERN and name of coordinator.

Question:

Please state the number of HCPs (CEs) participating in ERNs as full members (total across all ERNs), if known.

Question:

Please state the number of HCPs (CEs) participating in ERNs as associated/collaborating centres, if known.

Please proceed to next section

Question:

Please state the number of HCPs (CEs) participating in ERNs as associated/collaborating centres, if known.

Answer:

Free text comments box

Please proceed to next section

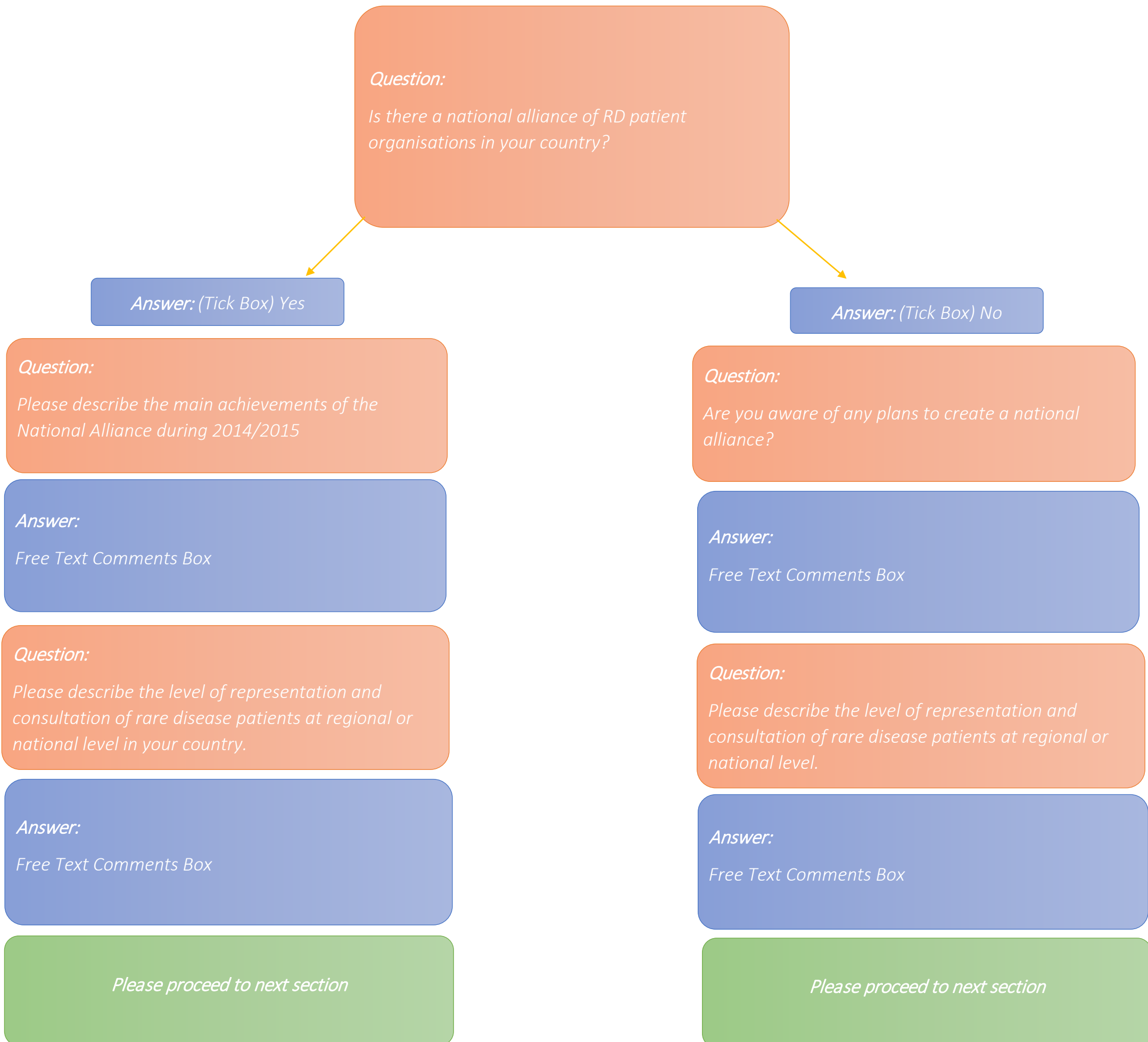
Answer:

Free Text to specify number

Answer:

Free Text to specify number

H. National Alliances of Patient Organisations and Patient Representations



I. National Helplines for RD

Question:

Is there a helpline in place dedicated to rare diseases in your country?

Answer: (Tick box) Yes

Answer: (Tick box) No

Question:

How is the helpline funded?

Answer: (Tick box)
PUBLIC Funding

Answer: (Tick box)
PRIVATE Funding

Answer: (Tick box)
Mixture of PUBLIC and PRIVATE Funding

Question:

Is the helpline available for: (select one)

Answer: (Tick box)
PATIENTS only

Answer: (Tick box)
PROFESSIONALS only

Answer: (Tick box)
PATIENTS/PROFESSIONAL (anyone)

Question:

Please briefly describe how the helpline(s) are operated.

Answer:

Free Text Comments box

Please proceed to next section

Question:

Please describe any plans to set up a helpline.

Answer:

Free Text Comments box

Please proceed to next section

J. Activities of Orphanet/Alternative RD-specific information system

Question:
Do you have an operational National Orphanet Team?

Answer. (Tick Box)
Yes

Answer. (Tick Box)
No

Question:
Please confirm the name of the institution currently hosting Orphanet.

Answer:
Free Text Comments Box

Question:
Please describe the activities and funding sources of any functional rare disease specific information system?

Answer:
Free Text Comments Box

Answer:
Please proceed to next section

Question:
Is this institution a partner in RD-Action?

Answer. (Tick Box)
Yes

Answer. (Tick Box)
No

Question:
Does this team produce information in your national language?

Answer:
Free Text Comments Box

Question:
Is there dedicated funding in your country's national plan or strategy for rare diseases to support the Orphanet team? (Is this funding provided nationally or regionally?)

Answer:
Free Text Comments Box

Question:

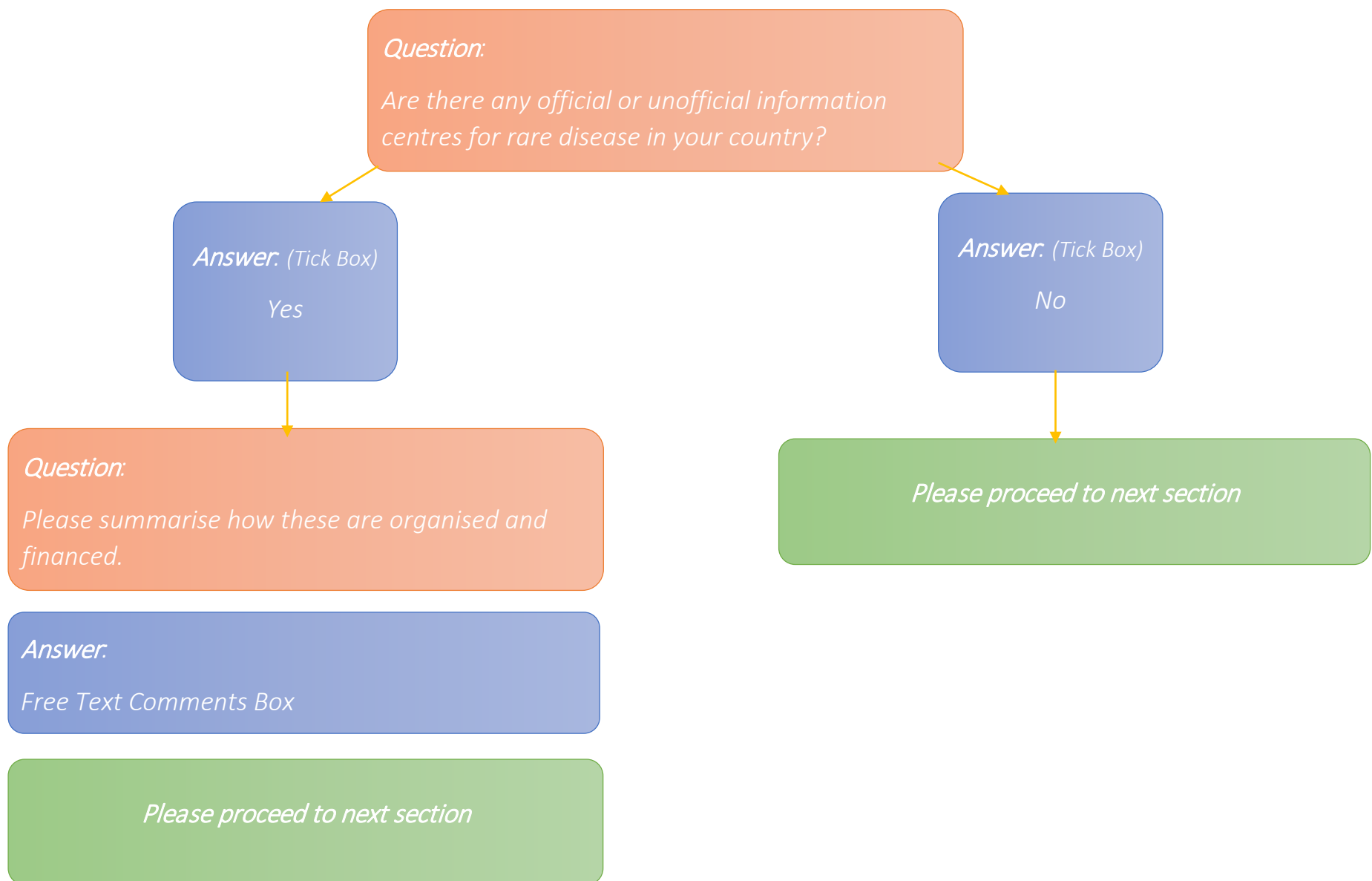
Please summarise the major activities of your national Orphanet team since the beginning of 2014.

Answer:

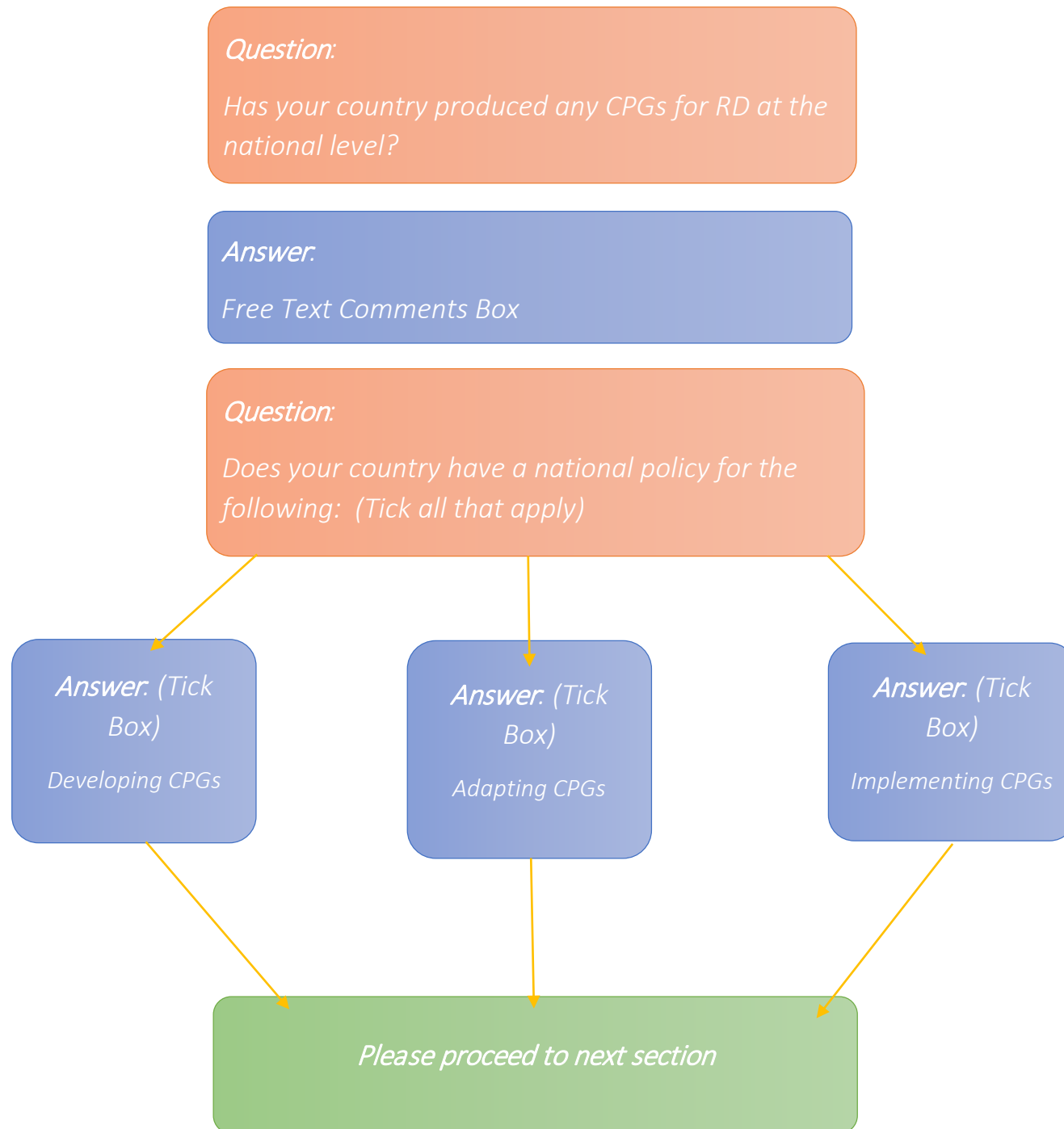
Free Text Comments Box

Please proceed to next section

K. Official Information Centres for Rare Diseases



L. Clinical Practice Guidelines (CPGs)



M. Training and Education

Question:

Please describe any initiatives in your country for RD related training/education.

Answer:

Free Text Comments Box

Please proceed to next section

N. Rare Disease Events

Question:

Please summarise any Rare Disease Day events which have taken place in your country since the beginning of 2014.

Answer:

Free Text Comments Box

Question:

Were there any other RD/OMP-focussed events organised by your country, either at the national or international level, since the beginning of 2014?

Answer:

Free Text Comments Box

Please proceed to next section

O. Existence of RD research programmes/projects in your country

Question:

Does your country have any specific programmes/projects to fund/facilitate rare disease research?

Answer: (tick Box)

Yes

Answer: (tick Box)

No

Question: (tick which best applies):

- a) There is a specific research programme for RD
- b) Specific Projects for RD are funded from the general research budget

Answer:

Free Text Comments Box for further details

Question:

Please summarise the status quo of rare disease research in your country (of all kinds – basic, clinical, translational, social/socio-economic).

Answer:

Free Text Comments Box

Question:

Has there been a policy decision to allocate a portion of the national research budget specifically to RD research?

Answer: (tick Box)

Yes

Answer: (tick Box)

In development

Answer: (tick Box)

No

Question:

Please stipulate total amount of public funds allocated for RD research actions/projects *per year* since the NP/NS began.

Answer:

Free Text Comments Box

Question:

Please summarise any ongoing/future plans to conduct social or socio-economic – focussed research into rare diseases.

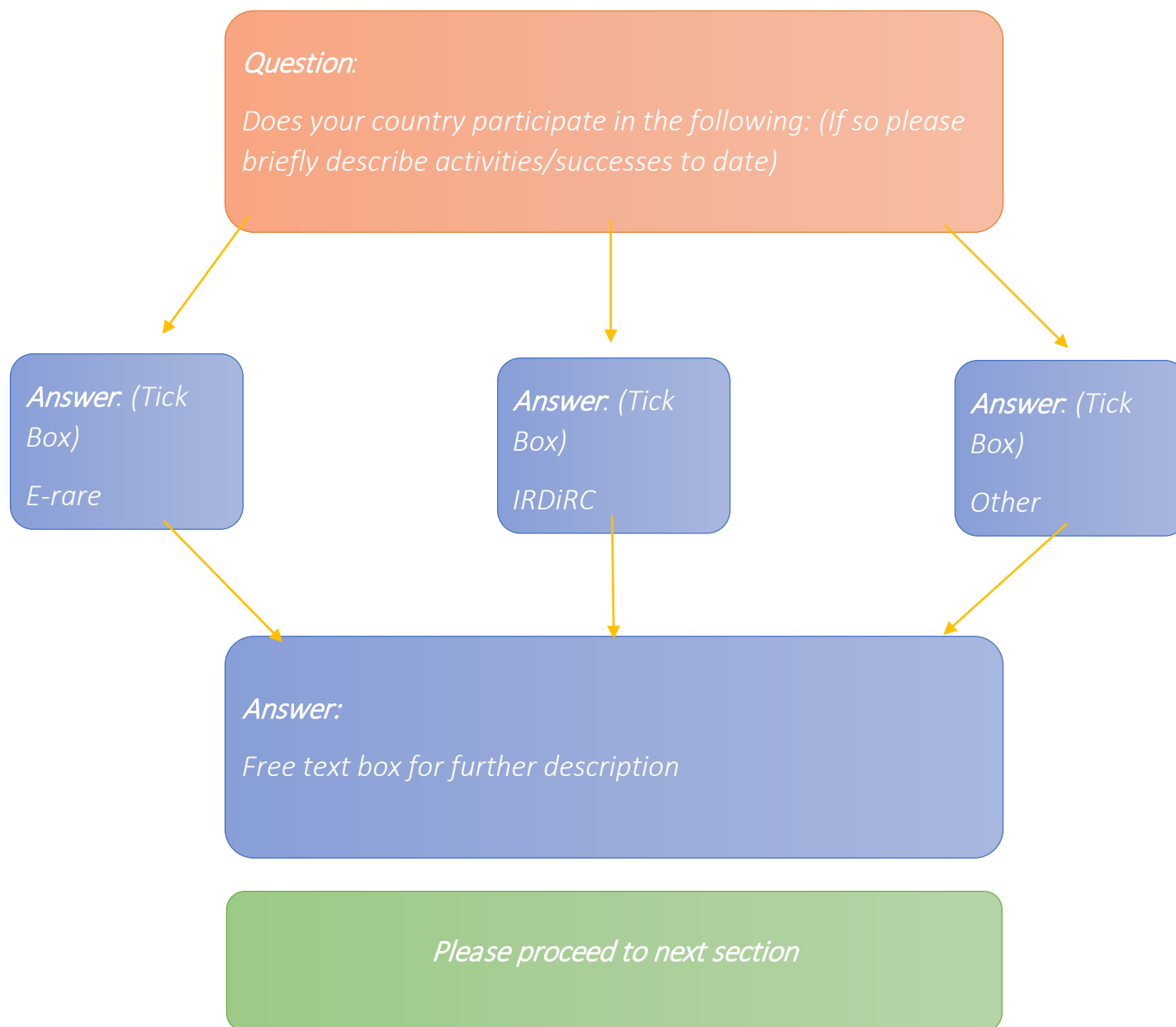
Answer:

Free Text Comments Box

Please proceed to next section

Please proceed to next section

P. Participation in E-rare and International Research Initiatives



Q. Orphan Medicinal Products (OMPs)

Question:

How many OMPs with a European Union marketing authorisation are available in your country (i.e. are priced and reimbursed or provided directly by your country's health system)? (And is this information available publicly in your country?)

Answer:

Free Text Comments Box

Question:

Does your country have any measures in place to facilitate access to Orphan Medicinal Products for patients?

Answer: (Tick Box)

Pre Authorisation Access (at a national level, not from industry or individuals): e.g. Compassionate use programmes; Cohort/named patient supply; expanded access programmes)

Answer: (Tick Box)

Specific approval procedures for pricing and reimbursement of **Orphan** Medicinal Products: e.g. Possibility of accelerated review; accelerated pricing process; specific expert groups/committees; reimbursement measures such as specific policies or restricted budget

Answer: (Tick Box)

Other measures for Orphan Drug Availability to Patients

Answer: Please provide details for each box you ticked

Free Text Comments Box

Question:

Does your country provide any incentives to support research* into, and the development of (designated or potential) Orphan Medicinal Products? (*research here encompasses anything from fundamental/basic to clinical development)

Answer: (Tick Waivers of Regulatory Fees)

Answer: (Tick Box)

Tax Incentives

Answer: (Tick Box)

Public Funding Schemes (schemes using funding from any Ministry, public agency, regional government body etc.)

Answer: (Tick Box)

Other types of measures to support development (e.g. free scientific advice from National Authority on drug development)

Please provide brief details of the Incentives you ticked:

Free Text Comments Box

Question:

Which of the following initiatives to support Orphan Drugs/Rare Diseases have been planned but have not yet been put in place in your country?

3 sections:

- 1. Measures to support research into Rare Diseases*
- 2. Measures to support development of Orphan Drugs*
- 3. Measures that support availability of Orphan Drugs to patients*

Section 1

Answer: (Tick Box)

- Research Programmes*
- Research Projects*
- Other Types of Measures*

Comments box: if known, please provide expected date of implementation

Section 2

Answer: (Tick Box)

- Fee waivers*
- Tax incentives*
- Public funding scheme*
- Other measures*

Comments box: if known, please provide expected date of implementation

Section 3

Answer: (Tick Box)

- Pre-authorisation*
- Specific approval procedures*
- Measures for post marketing authorisation obligations*
- Accelerated process of pricing*
- Reimbursement measures*
- Other measures*

Comments box: if known, please provide expected date of implementation

Please proceed to next section

R. Social Services and Social Integration

Question:

Which of the following best describes the existing programmes/services* in your country to support the integration of people with rare diseases into daily life activities?

*In particular

- a) Information and training for patients, family and caregivers (i.e. regarding their rights and available support measures/benefits);
- b) Daily life assistance for patients, family and carers (e.g. personal assistants, psychological support, respite care, therapeutic recreation programmes)
- c) Integration to employment for patients and family (e.g. incentives to hiring, protection in regards to absence from work, flexible schedule)
- d) Integration at school (e.g. individual support, training for teachers, good practices);
- e) Adapted housing (e.g. group homes, adapted housing grants)

Answer. (Tick Box)

Specific *programmes/facilities* exist to support people with rare diseases

Answer. (Tick Box)

Specific *actions* exist to enable real access for people with rare diseases to *general* social/disability programmes (i.e. training, guidelines for social workers etc.)

Answer.

Free Text Comments Box for further details

Question:

Are there any mechanisms to facilitate multidisciplinary, holistic, continuous care provision? (Tick all that apply)

Answer. (Tick Box)

Multidisciplinary/Inter-ministerial working group for rare disease, including representatives from social affairs/services Ministry (or equivalent)

Answer. (Tick Box)

Individual care plans including access to social and support services

Answer. (Tick Box)

Case manager services facilitating coordination of care and transfer of information between care providers.

Answer. (Tick Box)

Resource centres for RD – ‘one-stop shop style’ social services for RD.

Answer. (Tick Box)

Others – please indicate

Answer.

Free Text Comments Box for further details

Question:

Are there any specific measures to support the integration of rare diseases' specificities into the national system responsible for assessing a person's level of functioning (disability)?

Answer. (Tick Box)

YES

Answer. (Tick Box)

NO

Question:

Please outline any future plans to support social integration of people with RDs (i.e. in progress/development).

Answer:

Free text comments box for further details

Please proceed to next section

S. Other

Question:

Please provide details of any other RD related achievements in your country since the beginning of 2014.

Answer:

Free Text Comments Box

Question:

Please summarise what you see as the main areas of improvement/major successes relating to rare diseases since the beginning of 2014.

Answer:

Free Text Comments Box

End of Questions

Thank you for completing the questionnaire