Hi!

Here are some of the points I would like to make as a Finnish mother of a intellectually disabled rare disease child and patient group activist.

Question 1: Is the current EU definition of a rare disease satisfactory?

I would like the words "or rare syndrome" to be included in the definition of rare disease which I otherwise agree with.

Question 2: Do you agree that there is a pressing need to improve coding and classification in this area?

Yes. For example microdeletion syndromes are very individual and the diagnosis or classification may be based on known phenotype of an individual gene in the microdeletion area.

Question 4: Should the European Reference Networks privilege the transfer of knowledge? The mobility of patients? Both? How?

I would prefer one national center for rare disease or syndromes with congenital heart conditions. We are willing to travel to the capital from the north in order to see doctors with adequate expertise.

There are no specialists in the country for my child's condition. Nearest ones are in the UK but traveling there is too much to manage.

As a requirement for centers of reference for rare disease I have to insist that clear practical instructions are set that include for an example intellectually disabled,
non-verbal and severely disabled rare disease patients to get the same level of care as everyone else. Active control and measurement of discrimination in the health care "chain" also in the form of delay in receiving care must be required and results published EU wide.
Although this might seem unneccesary in light of real cases that have to my attention recently it is not given that doctors would not refuse blood tests, IV-fluids or referrals to specialists if the patient is severely disabled or intellectually disabled and often this discrimination is not recorded in the system at all.

Also hospitals that are selected a centers of reference should not have instructions or practices where severely disabled patients (seen as a group) would be issued DNRs against their or their families wishes and hence later denied intensive care and even IV-fluids and antibiotics. (This matter has been open for public discussion in Finland in the past year.)

Question 5: Should on-line and electronic tools be implemented in this area?

Electronic patient archives in connection with biobanks could be used

in research and providing relevant information to the currently undiagnosed rare disease patients.

Question 6: What can be done to further improve access to quality testing for RD?

Testing should always be accompanied by counseling. Birth of disabled or rare disease siblings should not be presented as a negative outcome.

Question 7: Do you see a major need in having an EU level assessment of potential population screening for RD?

No. It would be more practical for an example to have higher quality in ultrasound screening for congenital heart condition in the whole EU.

Question 8: Do you envisage the solution to the orphan drugs accessibility problem on a national scale or on an EU scale?

I feel that orphan drug reimbursement should be EU wide.
There should be a trial. Also we need
statistics on accessibility specifically in the disabled patient groups.

Question 9: Should the EU have an orphan regulation on medical devices and diagnostics?

A trial with a relevant medical devise would give more information.

Question 10: What kind of specialised social and educational services for RD patients and their families should be recommended at EU level and at national level?

Respite care should be offered with the option of an personal aid that aids the patient in participating the normal life with the family and friends - not to separate the patient from social contacts.

Information on rare disease should be produced and updated at EU level and translated into all official languages, nor recreated nationally many times. This way information facilitating patient mobility will not be lost by "modification to national conditions".

Patients should be supported in creating their own recreation groups in their own community. It is not practical for a hobby group to be limited by a diagnosis. Relevant peer contacts can be created by wider target group definitions such as "intellectually disabled or delayed children 2-7 years old with their siblings". Financial support for activities listed with the Eurordis Rapsody project would help and collecting best practices.

Question 11: What model of governance and of funding scheme would be appropriate for registries, databases and biobanks?

Finland in legislating biobanks and I agree with the proposal for the law that is available in Finnish.

Question 12: How do you see the role of partners (industry and charities) in an EU action on rare diseases?
What model would be the most appropriate?

All I know is that preferring funding models that require the patient groups to provide some of the research money will create more inequality as the most affected or disabled patients are not be able to work nor have such money.

Question 13: Do you agree with the idea of having action plans?
If yes should it be at national or regional level in your country?

Yes. National. I am very exited about the national plan for rare disease and feel that it is a real opportunity to integrate the disabled rare disease patients more fully in the highest level of care. And to apply the European ethical values.

In Finland the national plan for rare disease does not exist yet.
Also there is no umbrella organisation for rare disease patient groups.
Yet Eurordis has informed me of the European development and I am very grateful for all their hard work.

As individual co-operating patient groups in Finland we have put forward an initiative that there would be an entry point in the national orphan drug reimbursement process
(or a common drug used in an unusual way or for a rare diagnosis and hence is not covered in the reimbursement) where individual patients could easily request that the drug they need would be evaluated for reimbursement while now only pharmaceutical companies can apply for reimbursement or extension for reimbursement.
There is no trace in the system if a patient or a group with a rare diagnosis has to pay for a drug themselves or if due to disability or poverty they do not get the drug they need.

Question 14: Do you consider it necessary to establish a new European Agency on RD and to launch a feasibility study in 2009?

Yes. With it's primary task implemented in connection with applying disabled peoples human right and monitoring affects of discrimination and poverty.

Taina Hickey, Oulu, Finland

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