ERN services: A Patient Case Study

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Potential ERN Services Second Workshop, PWC, Brussels
24 May 2016
Collective knowledge and expertise

A European Reference Network is **more than the sum of its individual parts**!

- Services will include delivery of **specialist advice** on diagnostic, care and treatment, for rare and complex cases.
- Specialist advice will be based on ‘**collective experience, knowledge and expertise**’ generated in the network, which is more than the sum of its individual healthcare providers.
Method of delivery: specialist advice...

Direct method of the delivery of specialist advice service:
• Second opinion, virtual MDT or virtual tumour board to inform specialized care plans

Indirect method of generating specialist advice service:
• training, education, research and evidence generation, development of best practice, care standards, etc. …

Note: Delivery of specialist care is outside the ERN either through local providers or the Healthcare Provider.
Patient Case Framework

1. Proximity of doctor or emergency services
   Family doctor or a local medical center

2. Basic diagnostics
   Use of common diagnostics

3. Suspected diagnosis
   Due to low prevalence or lack of diagnostic means, doubts related to the diagnosis persist

4. Referral to specialist
   Could be a specialist doctor or a reference entity for the suspected issue

5. Further diagnostics
   Could be standard/usual diagnostic resource or a very specific one

6. Final diagnosis
   The suspected diagnostic is confirmed or a new diagnostic is given

7. Treatment
   Should follow clinical guidelines. For non-treatable conditions, a palliative plan will be established

8. Monitoring & Follow-up
   Preliminary results monitored. Self-monitoring could be used.

9. Relapse or worsening
   The treatment isn’t having the expected results or further issues arise

10. Updated Treatment
    The treatment is rethought based on non-achieved results and on new diagnosis. The treatment follows clinical guidelines.

11. Cure or Death
    The treatment cycle ends with the patient feeling well (cure or with palliative successful measures) or with their death
NF2 Patient Case Study
HCP specialising in Neurofibromatosis Type 2:

- HCP usually cover management of <500 patients or complex surgical procedures with <500/year
- HCP provided by limited number of centres (approx. 3-4 HCP in some Member States)
Neurofibromatosis Type 2: Summary

- Predisposes to benign tumours of the ‘linings’ of the nervous system
- Schwannomas on cranial, spinal root and peripheral nerves
- Meningioma and ependymomas
- Most frequent tumours are bilateral vestibular schwannomas (acoustic neuromas)
- Different approaches to treatment – surgery vs. radiosurgery for vestibular schwannomas

- Mean onset of symptoms 20 years (9-34 years)
- Mean survival WITHOUT surgery 18.5 years (4-44 years)
- Mean survival WITH surgery 9.2 (3-19 years)

Neurofibromatosis Type 2: Imaging
Treatment Pathway
1. Family doctor or emergency service

Description
- Anis developed pain and weakness in his legs at the age of ten.
- He was seen by his family doctor, who referred him to the HCP specialising in neurological cases.

ERN provide service
- Online sign posting of where the expert centres are in Germany.
- Cross-Border Healthcare Pathways

HCP resources usage
- Triage referral
Description:
• Investigations showed he had multiple lumbar spinal root tumours.
• He had a lumbar laminectomy and biopsy showed a schwannoma.

ERN services provided:
• Guidelines for diagnostics

Healthcare Provider Member resource usage:
• Hearing assessment
• MRI scan and diagnostic tests
• Neurological and genetic case review
• Lumbar laminectomy and biopsy
3. Suspected diagnosis

Description:
• Anis had increasing poor mobility and pain
• He was diagnosed with ‘?form of neurofibromatosis’

ERN services provided:
• Guidelines for diagnosis and onward treatment

Healthcare Provider Member resource usage:
• Genetic tests and counselling
• MRI and cranial scans
• MDT specialist review
4. Referral to specialist

Description:

- Anis was diagnosed with mosaic Neurofibromatosis type two.
- Mosaic patients only have the NF2 gene in some cells of their body, the mutation having occurred during their development. It is well recognised that in mosaic genetic conditions the area(s) of the body affected may be much more severely involved than usual.
- The main problem was pain that came from the schwannomas on the major nerves in his right leg.
- He was referred and assessed by a ERN virtual MDT specialising in NF2 due to severe spinal involvement in NF2 and schwannomas from T8 down.

ERN services provided:

- European evidence based clinical protocol is available.
- Triage patient referral – review
- Virtual MDT review
- Second opinion of complex case and specialist care plan given to HCP, including additional specific diagnostics required to be completed by HCP.

Healthcare Provider Member resource usage:

- Pain clinic assessment
- Prescribed combination of drugs for neuropathic pain and an opiate derivative.
5. Further diagnostics

Description:
• His neurological examination showed mild weakness in his right leg and sensory loss on the right lateral thigh. He was fully mobile and attending school regularly.

ERN services provided:
• Guidelines for diagnostics

Healthcare Provider Member resource usage:
• Additional specific diagnostics (e.g. phenotyping, new genes, array technique) completed by HCP.
• Neurological examination, MRI and cranial scan
6. Final diagnosis

Description:
- Molecular studies on blood and tumour confirmed a clinical diagnosis of probable mosaic NF2
- Prior to this Anis diagnosis had been ‘?form of neurofibromatosis’.

ERN services provided:
- Genetic tests reviewed and specialist advice given.

Healthcare Provider Member resource usage:
- Genetic test and molecular studies completed
- Clinical review
Description:
- He was assessed by neurosurgeons at the HCP specialist centre.
- Neurosurgeon felt that removal of the spinal root schwannomas was not possible without causing more neurological problems.

ERN services provided:
- Reference Best Practice of Diagnostic and Care, on which the treatment will be based.
- Capture data either as part of a protocol research for that disease or as part of a post-marketing real world evidence data collection.

Healthcare Provider Member resource usage:
- Neursurgery assessment completed, MRI scan and investigations.
- Surgery to remove larger right leg peripheral nerve schwannomas and this helped the leg pain.
Description:

- The power in his right leg began to deteriorate and for the first time since the laminectomy he developed weakness in the left leg.

ERN services provided:

- Data will be collected all along in the patient registration file which is a common tool across HCP in the ERN, collecting common data set, sharing patient summaries.
- ERN to ERN consultation of other more rare and less distinct clinical features and co-morbidities

Healthcare Provider Member resource usage:

- MDT follow up / monitoring
9. Relapse or worsening

Description:

- Weakness in his legs progressed resulting in Anis only being able to walk a few steps unaided.
- Repeat MRI scans showed rapid growth of the existing tumours and a new one slightly higher up which was already compressing the thoracic cord markedly.
- The mosaic nature of the disease was emphasised by his cranial scan which only showed slight growth in his known right trigeminal nerve and for the first time a tiny dot of enhancement on the right eight nerve suggestive of a vestibular schwannoma.
- There were no meningiomas which are common in non-mosaic severe NF2 presenting in childhood.
- Pain was again a major feature this time in the right flank which was thought to probably be due to the extra-spinal portion of a very large schwannoma pressing on the renal capsule.
- Once more the neurosurgeons felt they could not improve the situation.

ERN services provided:

- MDT virtual case review of clinical features and co-mobidities

Healthcare Provider Member resource usage:

- MRI scans, cranial scan
- MDT review, neurosurgery assessment and repeat scans
10. Updated treatment

Description:
• His weakness progressed resulting in Anis being unable to walk.
• He had no useful strength in the right leg, deteriorating weakness in the left leg and loss of sensation in both legs.
• His immobility meant that he was not eligible for Avastin as per the agreed national protocol.
• Advise on treatment based on exceptional nature of case from ERN.
• ERN felt that given the unusual nature of the patient’s disease and the fact that no other treatment offered any chance of improvement it was appropriate to start Avastin.
• Local commissioners approved treatment. Treatment to stop at one month if no radiographic or clinical response. He commenced 5mg/kg Avastin at fortnightly intervals

ERN services provided:
• MDT treatment planning and review to initiating of treatment (avastin)
• MDT follow up / monitoring (e.g. biomarker)
• Discharge clinical review

Healthcare Provider Member resource usage:
• Avastin treatment, monthly scans and monitoring of treatment
Description:

- Anis responded to treatment and was able to walk following treatment !!
- His one month scan showed a radiographic reduction in size and his lower limb neurology is almost normal after 8 doses.

ERN services provided:

- Discharge clinical review

Healthcare Provider Member resource usage:

- Avastin treatment continued, monthly scans and monitoring of treatment
- MDT follow up / monitoring (e.g. biomarker)
Cross-Border HealthCare pathways for patients in ERN, should include CBHC.

- identify the right HCP for this highly technical and complex expertise;
- prescription by the local HCP;
- recognition of this prescription by the National Contact Point;
- delivery of advice in the ERN:
  - preparation phase by the local HCP, and again the follow up by the local HCP
- delivery of treatment in the HCP:
  - complex intervention is delivered by another HCP in the ERN or another ERN
Holistic care

Holistic care for rare disease and co-morbidities.

Each new co-morbidity requires:

• new diagnosis
• new multiple discipline expertise
• possible expertise from doctors outside your country
• sharing of expertise within the ERN, etc
European Reference Networks‘:
• aim is to ‘improve access to high quality healthcare’
• ERN services are to inform treatment based on the ‘collective expertise’ of the network

Collective expertise opinion:
• Dependent on the level of expertise in each Member State
• Setting threshold for provision of ERN services would be different … defining threshold would be agreed MS by MS
• Second opinion was give prior to ERNs free at the good will of experts = cost was absorb by HCP
• With ERNs being established this formalises the sharing of expertise across Europe = cost will now need to be reimbursed
ERN services are …

• Indirect coordination services

• Direct clinical services
  – Triage patient referral – review
  – **Specialist care planning advice** to local / regional services for complex cases
  – MDT case review *and with additional specific diagnostics (e.g. phenotyping, new genes, array technique)*
  – Highly specialised surgery or intervention in HCP
  – MDT treatment planning and review to initiating of appropriate treatment
  – MDT follow up / monitoring (e.g. biomarker)
  – Discharge clinical review

• **Knowledge generation and dissemination**
  – **Clinical audit events** for sharing and dissemination of knowledge, evidence and expertise and identify emerging best practice
  – Development of clinical / best practice guidelines, referral pathways
  – Teaching, training and continuous education events for ERN’s HCP and externally for local, regional and national healthcare providers
  – Discussion and learning through eForums

* Additional tests and treatments provided in HCP not ERN
Thank you

Daniel - Sanfilippo syndrome