Current status of National Intractable Disease (Nambyo) registry in Japan. - History, current issues, new trials, and future directions -

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What is Intractable Disease

• In 1972, the Ministry of Health and Welfare enacted the “General Outlines for taking Measures to deal with Intractable Disease [NAMBYO]” which was first established in the world. The extent of Nambyo was proposed as
  – Unknown etiology
  – Un-established therapy
  – Frequent after-effects
  – Chronic course
  – Heavy burdens in terms of economical, psychological and physical issues.

• At this stage, there was no concept of rareness in the extent of Nambyo. More than 25 years was needed until rareness was added to the extent.
Revised definition of Nambyo in 1995.

- Etiology is unknown.
- Frequency is low (less than 50,000).
- Therapy is not established.
- Economical, psychological and physical burdens.
- Diagnostic criteria should be established.
The present status of Nambyo.

• Selection of Nambyo
  – 130 diseases were selected as Nambyo for special research.
  – 214 diseases were further selected as candidates of the second group of Nambyo for research support.
  – Of 130 diseases, 56 diseases are targets for being treated with special favor in terms of medical expenses.

• Size of budgets
  – Budgets for researches (364) 10 billion yen
  – Budgets for medical expenses (56) 35 billion yen

• Number of Recipients of Medical Expenses
  – Approximately 700,000 (increased by 30,000/y)
The movement of Number of Recipients of Medical Expenses

700 thousand patients (2010.3)

Ulcerative Colitis
Parkinson's disease
Systemic Lupus Erythematosus
Problems of “NAMBYO”

• Comprehensive measures for Nambyo is important
• Development of drugs for Nambyo is not proceeding
• Providing information about Nambyo should be promoted even more
• Issues for medical expense support
List of 56 Disease for Registry

01 Behcet disease
02 multiple sclerosis
03 myasthenia gravis 0
04 systemic lupus erythematosus (SLE)
05 subacute myelo-optico-neuropathy (SMON)
06 aplastic anemia
07 sarcoidosis
08 amyotrophic lateral sclerosis (ALS)
09 scleroderma, dermatomyositis, or polymyositis
10 idiopathic thrombocytopenic purpura
11 (1) polyarteritis nodosa  (2) microscopic polyangiitis
12 ulcerative colitis
13 Takayasu arteritis
14 thromboangiitis obliterans, Buerger disease
15 pemphigus
16 spinocerebellar degeneration
17 Crohn disease
18 fulminating hepatitis
19 malignant rheumatoid arthritis (rheumatoid vasculitis)
20 Parkinson disease and related diseases  - progressive supranuclear palsy  - corticobasal degeneration  - Parkinson disease
21 amyloidosis
22 ossification of posterior longitudinal ligament (OPLL)
23 Huntington disease
24 moyamoya disease
25 Wegener granulomatosis
26 dilated cardiomyopathy, congestive cardiomyopathy
27 multiple system atrophy  - striatonigral degeneration (SND)  - olivopontocerebellar atrophy (OPCA)  - Shy-Drager syndrome
28 epidermolysis bullosa
29 pustular psoriasis
30 disseminated spinal canal stenosis
31 primary biliary cirrhosis
32 severe acute pancreatitis
33 idiopathic necrosis of the femoral head
34 mixed connective-tissue disease
35 primary immunodeficiency syndrome
36 idiopathic interstitial pneumonia
37 retinitis pigmentosa
38 prion diseases  - Creutzfeldt-Jakob disease (CJD)  - Gerstmann-Straussler  - Sheinker syndrome  - fatal familial insomnia
39 primary pulmonary hypertension
40 neurofibromatosis type 1, neurofibromatosis type 2
41 subacute sclerosing panencephalitis (SSPE)
42 Budd-Chiari syndrome
43 idiopathic chronic pulmonary thromboembolism with pulmonary hypertension
44 lysosomal storage diseases  - Fabry disease  - other lysosomal storage diseases
45 adrenoleukodystrophy (ALD)
46 Familial Hypercholesterolemia ( homozygous type)
47 spinal muscular atrophy (SMA)
48 spinal and bulbar muscular atrophy (SBMA)
49 Chronic Inflammatory Demyelinating Polyneuropathy
50 Hypertrophic cardiomyopathy
51 Restrictive cardiomyopathy
52 Mitochondrial disease
53 lymphangioleiomyomatosis
54 Severe erythema multiforme (Acute Phase)
55 Ossification of the ligamentum flavum
56 Diencephalo-hypophysial dysfunction  - Syndrome of abnormal secretion of prolactin  - Syndrome of abnormal secretion of gonadotropin  - Syndrome of abnormal secretion of antidiuretic hormone  - Syndrome of abnormal secretion of Thyroid stimulating hormone  - Cushing disease  - Acromegaly  - Hypopituitarism

*The diseases with red color are implemented in this study.
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Future measures of Nambyo

• Overall reviewing of measures of Nambyo is currently carried out:
  – Medical system
  – Subsidized health care costs
  – Promotion of research
  – Consultation and support
  – Human services
  – Work and employment support
  – International cooperation
  – Awareness
Current status of reviewing future measures of Nambyo

• Interim Report of the Committee for Measures of Intractable Diseases in December 2011

• Cabinet decision of Measures for Intractable diseases in the future in February 2012
  – Aims to construct a mechanism to implement comprehensive measures and support for clinical research, medical care, welfare services, and employment support.
  – Aims to develop a mechanism to support medical expenses, considering legislation.
Problems on data repository

• Certificate criteria
  – Vary from prefecture to prefecture

• National data registry
  – Nationwide follow up system
  – Quality management
  – Epidemiological new findings
  – Security issues
  – Lacks of semantic interoperability
MindMap current version
We have modeled 49 archetypes for six intractable diseases

- New fifteen archetypes
- 34 specialized archetypes from CKM

Templates of intractable disease use many archetypes:

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# List of archetypes

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N: new  S: specialized
Japan Specific archetypes

- Originally designed for health insurance of Japan.
- The healthcare system widely differs from one country to another.
Specialized Diagnosis

- Specialized from problems-diagnosis for describing severity in Parkinson’s disease

Differences in severity index

Differences in severity description
Originally created archetypes

Specialized Medical Treatments not covered in CKM

Expanded Disability Status Scale

- Pyramidal
- Cerebellar
- Brain Stem
- Sensory
- Bowel and bladder functions
- Visual
- Mental
- Other