Common Genetic Problems in Internal Medicine

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A 15-YEAR OLD MAN PRESENTED WITH SHORT STATURE, OBESITY AND HYPOGONADISM
Ht. 145 cm, Wt. 72 kg, undescended left, testis Tanner stage 1, Karyotype 46, XY

Q1: What is the most appropriate investigation to obtain the diagnosis
A. FISH for 22q11
B. SNRPN methylation study
C. Parathyroid hormone level
D. PCR for SRY
E. Leptin level
Definition of Short Stature

- Height lower than 3rd centile in growth charts (<=-2sd)
- Severe short stature: Height lower than 1st centile (< -3sd)

-2SD is approximately 145 cm in Thai female and 158 cm in Thai male
### Height of Thai Medical Students and Residents Age 20-35 years (cm)

<table>
<thead>
<tr>
<th>Sex</th>
<th>P3</th>
<th>P25</th>
<th>P50</th>
<th>P75</th>
<th>P97</th>
</tr>
</thead>
<tbody>
<tr>
<td>M</td>
<td>165.91</td>
<td>170.25</td>
<td>174.00</td>
<td>178.00</td>
<td>185.00</td>
</tr>
<tr>
<td>F</td>
<td>150.73</td>
<td>156.75</td>
<td>160.00</td>
<td>163.00</td>
<td>168.00</td>
</tr>
</tbody>
</table>

Tanprom and Trachoo, Rama Med J 2015
Target height

Girls (cm) = $\frac{\text{mat height}+(\text{pat height}-13)}{2}$

Boys (cm) = $\frac{\text{pat height}+(\text{mat height}+13)}{2}$
Short Stature

**Physiologic**
- Familial
- Normal variant
- Constitutional delay
  - Chronic disease

**Pathologic**
- Proportionate
  - Malnutrition
  - Drug
  - Endocrine
- Genetics
  - Chromosome
    - Single gene
  - Skeletal dysplasia
    - MPS 4
    - Spine deformities
- Disproportionate
  - Short limb
  - Short trunk

Trachoo O., *in press*
# Table 1. Differential diagnosis of short stature and growth failure

<table>
<thead>
<tr>
<th>Healthy but short children</th>
<th>Endocrinopathies</th>
</tr>
</thead>
<tbody>
<tr>
<td>familial short stature</td>
<td>hypothyroidism</td>
</tr>
<tr>
<td>constitutional growth delay</td>
<td>hypopituitarism</td>
</tr>
<tr>
<td><strong>Intrinsic short stature</strong></td>
<td>heredity, sporadic, idiopathic</td>
</tr>
<tr>
<td>small for gestational age</td>
<td>isolated GH deficiency</td>
</tr>
<tr>
<td>genetic syndromes</td>
<td>birth injury</td>
</tr>
<tr>
<td>Down syndrome, Turner syndrome</td>
<td>craniopharyngioma</td>
</tr>
<tr>
<td>Prader-Willi syndrome</td>
<td>cranial irradiation</td>
</tr>
<tr>
<td>skeletal dysplasia</td>
<td>brain tumours</td>
</tr>
<tr>
<td>achondroplasia, hypochondroplasia</td>
<td>midline defects</td>
</tr>
<tr>
<td><strong>Systemic diseases</strong></td>
<td>haemosiderosis</td>
</tr>
<tr>
<td>infectious</td>
<td>GH insensitivity (Laron syndrome)</td>
</tr>
<tr>
<td>HIV, tuberculosis</td>
<td>glucocorticoid excess</td>
</tr>
<tr>
<td>cardiac disease</td>
<td>Cushing syndrome, exogenous steroids</td>
</tr>
<tr>
<td>renal disease</td>
<td>poorly controlled diabetes mellitus</td>
</tr>
<tr>
<td>renal tubular acidosis</td>
<td>precocious puberty</td>
</tr>
<tr>
<td>chronic renal insufficiency</td>
<td>pseudohypoparathyroidism</td>
</tr>
<tr>
<td>gastrointestinal</td>
<td>pseudopseudohypoparathyroidism</td>
</tr>
<tr>
<td>cystic fibrosis</td>
<td><strong>Nonorganic aetiologies</strong></td>
</tr>
<tr>
<td>inflammatory bowel disease</td>
<td>psychosocial deprivation</td>
</tr>
<tr>
<td>central nervous system disease</td>
<td>nutritional dwarfing</td>
</tr>
<tr>
<td>chronic lung disease</td>
<td></td>
</tr>
<tr>
<td>malignancy</td>
<td></td>
</tr>
</tbody>
</table>

*Abbreviation. GH, Growth hormone*
<table>
<thead>
<tr>
<th>Table 1. Differential diagnosis of short stature and growth failure</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>History</strong></td>
</tr>
<tr>
<td>Antenatal</td>
</tr>
<tr>
<td>complications of pregnancy,</td>
</tr>
<tr>
<td>pre-eclampsia, hypertension</td>
</tr>
<tr>
<td>maternal history of smoking, alcohol, infections</td>
</tr>
<tr>
<td>Birth</td>
</tr>
<tr>
<td>gestational age</td>
</tr>
<tr>
<td>birth weight and length</td>
</tr>
<tr>
<td>mode of delivery (breech, forceps)</td>
</tr>
<tr>
<td>Apgar score</td>
</tr>
<tr>
<td>neonatal complications</td>
</tr>
<tr>
<td>Nutrition</td>
</tr>
<tr>
<td>general well being: appetite, energy, sleep,</td>
</tr>
<tr>
<td>and bowel habits.</td>
</tr>
<tr>
<td>pattern of growth from birth</td>
</tr>
<tr>
<td>Developmental milestones</td>
</tr>
<tr>
<td>Maternal and child relationship</td>
</tr>
<tr>
<td>Medical history</td>
</tr>
<tr>
<td>underlying illness, drug intake, irradiation</td>
</tr>
<tr>
<td>Family History</td>
</tr>
<tr>
<td>short stature (3 generations).</td>
</tr>
<tr>
<td>age of onset of puberty in family members of the same sex.</td>
</tr>
<tr>
<td>diseases in the family.</td>
</tr>
</tbody>
</table>
Approach to Obesity

- Essential obesity
- Drug-induced: insulin, steroid, anti-depressant, etc.
- Endocrine: hypothyroidism, Cushing’s
- Genetics
  - Syndromic
  - Non-syndromic or monogenic obesity, e.g., familial leptin deficiency
Prader-Willi syndrome

- Microdeletion on chromosome 15
- Uniparental disomy
- Methylation defects
- Associated with advance paternal age
Signs of Prader-Willi syndrome may be seen at birth.
- Newborns are often small and floppy.
- Male infants may have undescended testicles.
- Trouble eating as an infant, with poor weight gain
- Almond-shaped eyes
- Delayed motor development

Narrow bifrontal skull
- Rapid weight gain during childhood
- Search for food and tempers tantrum
- Short stature
- Slow mental development
- Very small hands and feet in comparison to the child's body
- Hypogonadism
PWS - systemic complications

- Hypertension
- Diabetes mellitus
- Dyslipidemia
- Metabolic syndrome
- Sleep apnea
- Right-sided heart failure
- Orthopedic problems, e.g., osteoarthritis
Narrow temple distance and nasal bridge

Almond-shaped eyes
Mild strabismus

Thin upper lip
Downturned mouth

Overweight
Genetic testing and counseling of recurrent risk

Prader-Willi syndrome

- **Normal**: pat, mat
- **De novo deletion**: pat, mat
- **Maternal uniparental disomy**: mat, mat
- **Imprinting defect**: mat, pat

<table>
<thead>
<tr>
<th></th>
<th>Frequency</th>
<th>Recurrence risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal</td>
<td>~ 70%</td>
<td>&lt; 1%</td>
</tr>
<tr>
<td>De novo deletion</td>
<td>25 - 30%</td>
<td>&lt; 1%</td>
</tr>
<tr>
<td>Maternal UPD</td>
<td>~ 1%</td>
<td>&lt; 1%</td>
</tr>
<tr>
<td>Imprinting defect</td>
<td>no IC deletion</td>
<td>50% IC deletion</td>
</tr>
</tbody>
</table>
Turner’s Syndrome

- 45 XO: most common karyotype
- Mosaicism with 46, XX and/or 47, XXX commonly found
- Proportionate short stature
- Webbed neck or skin redundancy of the neck
- Low posterior hairline
- High-arch palate
- Cubitus valgus
- Shortening 4th and 5th metacarpal or metatarsal bone
Turner’s Syndrome

• Coarctation of aorta (17-45%)
• ASD, VSD
• Hypogonadism, horse-shoe kidney, lymphedema
• Shield chest
• Widening nipple
• Occasionally low-set ear, epicanthal fold
• Intelligence- usually normal
• DDx- Noonan’s syndrome, Pituitary dwarfism (Combined pituitary hormone deficiency syndrome), Albright hereditary osteodystrophy
Obesity, short stature, learning disability, hypocalcemia
Albright’s Hereditary Osteodystrophy

- “Pseudohypoparathyroidism”
- Obesity, round face, shortening 4th and 5th metacarpal bones
- Mild degree of developmental delay
- GNAS1 mutation
- Autosomal dominant inheritance
- Paternal allele expressed resulting in Pseudopseudohypoparathyroidism, only allele inherited from mother being fully expressed “Genomic Imprinting”
Short stature, dysmorphic features, pulmonic stenosis
Noonan Syndrome

- Autosomal dominant
- Genetic heterogeneity
- Short stature
- Congenital heart defect: PS, HCM
- Developmental delay
- Broad or webbed neck
- Pectus deformities
- Apparently low-set nipples

- Cryptorchidism
- Characteristic facies: low-set ears with posteriorly rotated helices, wide-spaced eyes, epicanthal folds, thick or droopy eyelids
- Others: coagulation defects, platelet dysfunction, lymphatic dysplasias
Achondroplasia

- Most common cause of disproportionate short stature in children and adults
- Prevalence 1/27000, FGFR3 mutation (G1138A and G1138C)
- Autosomal dominant inheritance, 80% de novo
- Short limbs, normal-size trunk
- Male 120-145 cm, female 115-137 cm
- Complications: cervicomedullary compression, sleep apnea, spinal stenosis
Achondroplasia

- Disproportionate short stature
- Limb shortening predominantly rhizomelic with tibial bowing
- Wedge-shaped gap between the 3rd and 4th fingers -> “trident hand”
- Midface hypoplasia, depressed nasal bridge, large jaw, prominent forehead
- Limitation of elbow extension
- Evidence of tonsillar or adenoid enlargement
- Gait abnormalities due to shape of pelvis and spinal cord claudication
Achondroplasia: complications

- Anesthetic risks
- C-spine stenosis
- Glue ear
- Growth
- Hydrocephalus
- Pregnancy
- Hypermobility joints
- Respiratory insufficiency/OSA
- Spinal stenosis
Down syndrome

- Trisomy 21, translocation, mosaicism
- Risk in pregnant woman < 35 years = 1:600, >35 years, starting from 1:270
- Most common single cause of human birth defects
Down syndrome - physical signs

- Decreased muscle tone at birth
- Excess skin at the nape of the neck
- Flattened nose
- Separated joints between the bones of the skull (sutures)
- Single crease in the palm of the hand
- Small ears
- Small mouth
- Upward slanting eyes
- Wide, short hands with short fingers
- White spots on the colored part of the eye (Brushfield spots)
Down Syndrome

Features

- Upslanting palpebral fissure
- Brachycephaly
- Flat nasal bridge, hypoplastic maxilla
- Microtic, low-set ears
- Brushfield spots
- Sandal deformity
- Simian crease
- Excessive nuchal folds
- Epicantus
- Macroglossia, glossoptosis
Down syndrome - mental problems

- Impulsive behavior
- Poor judgement
- Short attention span
- Slow learning
Down syndrome - medical conditions

- Congenital heart defects: perimembranous VSD, ASD
- Dementia
- Cataracts
- Ear infections
- Hip dislocation
- Chronic constipation
- Sleep apnea
- Chewing problem
- Hypothyroidism
- Diabetes and metabolic syndrome
- Infantile GI tract blockage such as duodenal or esophageal atresia
Short stature, dysmorphic features, mental retardation, hypocalcemia, low PTH
22q11.2 deletion syndrome

- DiGeorge Syndrome
- Velocardiofacial Syndrome
- Shprintzen Syndrome
- Conotruncal Anomaly Face Syndrome (CTAF)
- Caylor Cardiofacial Syndrome
- Autosomal Dominant Opitz G/BBB Syndrome
- Deletion 22q11 syndrome
22q11.2 deletion syndrome - clinical manifestations

- Congenital heart disease (particularly conotruncal malformations)
- Palatal abnormalities [especially velopharyngeal insufficiency (VPI)]
- Hypocalcemia due to primary hypoparathyroidism
- Immune deficiency
- Learning difficulties
- Characteristic facial features
22q11.2 syndrome - clinical manifestations

- **Skeletal findings**: pre and postaxial polydactyly of the hands and postaxial polydactyly of the feet, supernumerary ribs, hemivertebrae, craniosynostosis

- **Genitourinary tract anomalies**: renal agenesis, hydronephrosis, multicystic/dysplastic kidneys, duplicated kidney, horseshoe kidney, absent uterus, hypospadias, inguinal hernia, and cryptorchidism

- **Laryngotracheal-esophageal abnormalities**: vascular ring and laryngeal webs

- **Ophthalmologic findings**: tortuous retinal vessels, ptosis, posterior embryotoxin, scleracornea, coloboma, cataract, and strabismus

- **CNS abnormalities**: cerebellar atrophy, polymicrogyria, enlarged sylvian fissures, neural tube defects, tethered cord, unprovoked seizures, and asymmetric crying facies

- **Gastrointestinal anomalies**: anteriorly placed anus or imperforate anus, esophageal atresia, jejunal atresia, accessory spleens, umbilical hernia, diaphragmatic hernia, intestinal malrotation, and Hirschsprung disease

- **Preauricular tags**
Less frequent seen manifestations

- Severe dysphagia
- Growth hormone deficiency
- Autoimmune disease (thrombocytopenia, juvenile rheumatoid arthritis, Grave's disease, vitiligo, neutropenia, hemolytic anemia)
- Hearing loss (sensorineural and conductive)
- Psychiatric illness
- Tumor: hepatoblastoma, renal cell carcinoma, Wilm’s tumor, neuroblastoma
22q11.2- cardiac complication

- Tetralogy of Fallot (TOF) 22%
- Interrupted aortic arch (IAA) 15%
- Ventricular septal defect (VSD) 13%
- Truncus arteriosus (TA) 7%
- Vascular ring 5%
- Atrial septal defect 3%
- Aortic arch anomaly 3%
- VSD; ASD 4%
- Other 4%
- Normal 26%
A 16-YEAR OLD THAI BOY PRESENTED WITH PNEUMOHEMOTHORAX
History

ชายไทยอายุ 16 ปี นักเรียน ม.5

CC: เจ็บแน่นหน้าอก 1 วัน

PI: 1 วัน ขณะเดินขึ้นบันได เจ็บหน้าอกแผลปุบๆ ทำทางมด้านซ้าย รู้สึกเหมือนได้ยินเสียงลูกโป่งไปยังหน้าอก ปวดร้าวแขนซ้าย หายใจ เข้าจดมากขึ้น มา ER ตรวจร่างกายพบ decrease breath sounds with tympanic percussion over left lung.
ไม่มีประวัติครอบครัวเรื่องโรคหัวใจ โรคปอด หรือเสียชีวิตแบบฉับพลัน
Pneumothorax of left lung
Thumb’s sign  Wrist’s sign  Pes planus
Long armspan  Arachnodactyly and joint hypermobility
Q2: What is the most likely diagnosis of his clinical features?

A. Physiologic tall stature
B. Marfanoid appearance
C. Marfan syndrome
D. Ehlers-Danlos syndrome
E. Homocystinuria
Tall stature

- Familial tall stature
- Excessive calorie intake/multifactorial
- Endocrine: pituitary gigantism
- Genetic disorders
  - Marfanoid
  - Eunuchoid
  - XYY
- Overgrowth disorders
Marfan Syndrome (MFS)

- Genetic disorder affecting the body’s connective tissue
- Prevalence 1:5000
- Mutations in FBN1, encoding fibrillin-1
- Subsequent increase of transforming growth factor beta (TFG-β)
- Autosomal dominant inheritance, 75% inherited, 25% de novo
- Clinical features commonly found in aorta, heart, bones, joints ad eyes
Dolichocephaly

Pectus excavatum

Pectus carinatum
High-arch palate

Kyphoscoliosis
Arachnodactyly

Thumb’s sign

Wrist’s sign
Striae atrophicae

Normal striae
Reduced elbow extension

Recurrent inguinal/incisional hernia
Aortic root dilatation > 2SD (from sinus of volsava) or Z score > 2
Aortic regurgitation
Mitral valve prolapse
Mitral regurgitation
Apical lung bleb with or without pneumothorax

Severe myopia and ectopia lentis (iridodonesis)

Lumbosacral dural ectasia
Cardiac Manifestations in MFS

- Dilatation of ascending aorta $Z$ score $\geq 2$ at least involving the sinus of vallsva (with or without aortic regurgitation)
- Dissection of ascending aorta
- Mitral valve prolapse with or without regurgitation
Figure: Aortic root growth curves normalised to body size and age

The left graph is for children and adolescents, the middle graph is for adults younger than 40 years, and the right graph is for adults older than 40 years. The shaded areas represent the range of values in which 95% of the population will fall. Modified from The American Journal of Cardiology with permission of Elsevier.39
Other Cardiac Features That May Be Seen

- Dilatation of main pulmonary artery
- Calcification of mitral annulus
- Primary dissection and/or dilatation of descending thoracic or abdominal aorta (approximately 10% of MFS population)
- Tricuspid valve prolapse
Revised Ghent Nosology Criteria 2010 for MFS in an adult > 20 years old*

• In the absence of family history
  - Ao (Z ≥ 2) + EL
  - Ao (Z ≥ 2) + FBN1 mutation
  - Ao (Z ≥ 2) + Systemic (≥ 7 pts)
  - EL + FBN1 mutation**

*Age < 20, Ao is estimated at Z ≥ 3
**Known causative mutation for Ao

Ao = aortic root dilatation
EL = ectopia lentis
FH = family history
Revised Ghent Nosology Criteria 2010 for MFS in an adult

- In the presence of family history
  - FH + EL
  - FH + Ao ($Z \geq 2$)
  - FH + Systemic ($\geq 7$ pts)

Ao = aortic root dilatation
EL = ectopia lentis
FH = family history
Systemic Scores

- Wrist and thumb’s sign 3
- Pectus carinatum 2
- Hindfoot deformity 2
- Spontaneous pneumothorax 2
- Dural ectasia 2
- Protrusio acetabulae 2
Systemic Scores

- Wrist or thumb’s sign 1
- Pectus excavatum/chest asymmetry 1
- Pes planus 1
- Armspan:height > 1.05 or U:L < 0.86 1
- Scoliosis > 20 degree/TL kyphosis 1
- Reduced elbow extension<170 degree 1
- Facial features 1
- Striae atrophicae 1
- Myopia 1
- Mitral valve prolapse 1
Z-SCORE CALCULATION

Aortic Root Z-Scores for Children

For patients up to 25 years of age utilizing systolic, inner to inner edge measurement of the sinuses of Valsalva according to Colan SD et al. J Am Coll Cardiol 2005;47:1858-65

- Height (cm)
- Weight (kg)
- BSA

Ao Root at sinuses of Valsalva (in cm)

Calculation:

Z-Score: 0

Closing remarks:


Differential diagnosis of Marfanoid appearance

- Marfan syndrome and related disorders
- Homocystinuria
- Klinefelter syndrome
- Fragile X syndrome
- Multiple endocrine neoplasia 2B
MASS Phenotype

Minor criteria for MFS

M (Myopia, MVP)
A (Aortic root dilate, minor)
S (Skeletal, minor)
S (Skin involvement)

or Ao Z<2 + Systemic score from 5 onwards
Hypermobility Joint Syndrome

- Other syndrome with internal organ complication to be excluded
- Normal life style and expectancy
- Aware of early osteoarthritis
- Genetic heterogenity
Hypermobility Joint Syndrome
Q3: What is the most likely diagnosis if echocardiogram and eye exam are normal?

A. Physiologic tall stature
B. Marfanoid appearance
C. Marfan syndrome
D. Marfan syndrome not excluded, follow-up is required
E. Please work up chromosome to exclude Klinefelter syndrome
Marfanoid appearance with chest pain

- Aortic dissection
- Pneumothorax
- Acute coronary syndrome
- Musculoskeletal pain
- Functional
Marfanoid appearance with back pain

- Spondylolisthesis
- Musculoskeletal pain
- Aortic dissection
- Lumbosacral dural ectasia
Marfanoid appearance with headache

- Venous sinus thrombosis due to homocysteinuria
- Intracranial hypotension due to CSF leakage at dural ectasia
- Functional
Klinefelter syndrome (47, XXY)

- 1:500 - 1:1000
- Tall stature as eunuchoid appearance (long legs, short trunk, shoulder equal to hip size)
- Gynecomastia
- Infertility
- Hair less than normal
- Sexual problem
- Small and firm testicles
Klinefelter syndrome - systemic involvement

- Enlarged teeth with a thinning surface ‘Taurodontism’
- ADHD
- Autoimmune diseases, e.g., SLE, RA, Sjogren
- Breast cancer
- Depression
- Learning disabilities, i.e., dyslexia
- Extragonadal germ cell tumor
- Osteoporosis
- Varicose vein
Taurodontism
48, XYYY (Klinefelter variant)
Triple X syndrome (47, XXX)

- 1:1000
- Tall stature
- Microcephaly
- Epicanthal folds
- Learning disabilities
- Delayed language and motor skills
- Low self-esteem and anxiety
- Commonly normal fertility
Angiofibroma

Ash leaf

Shagreen patch

Periungual fibroma s/p excision
Tuberous Sclerosis

- Autosomal dominant inheritance
- \textit{TSC1} and \textit{TSC2} gene on chromosome 9 and 16, respectively
- Skin involvement: adenoma sebaceum, Ash leaf, Shagreen patch, Confetti spot
- Neurological involvement: seizure, mental deficiency
- Pulmonary involvement: lymphangiomymoma
Tuberous Sclerosis

• Cardiac involvement: rhabdomyoma
• Multiple tumor: abdominal angiomyolipoma, ependymoma, astrocytoma
• Serious complications: bleeding tumor, pneumothorax
Common presentation of TSC in Internal Medicine

- Ruptured AML
- LAM with pneumothorax
- Seizure disorder
Neurofibromatosis Type 1

- Autosomal dominant inheritance
- NF1 gene on chromosome 17
- Skin involvement: café au lait spot, neurofibroma, axillary and groin freckling, plexiform neurofibromatosis
- Ocular involvement: optic glioma, Lisch’s nodule
- Bone and joint involvement: pseudoarthrosis, scoliosis, spondylolisthesis
- Learning difficulties
Clinical criteria: two or more

- Six or more café au lait macules over 5 mm in greatest diameter in prepubertal individuals and over 15 mm in greatest diameter in postpubertal individuals
- Two or more neurofibromas of any type or one plexiform neurofibroma
- Freckling in the axillary or inguinal regions
- Optic glioma
- Two or more Lisch nodules (iris hamartomas)
- A distinctive osseous lesion such as sphenoid dysplasia or tibial pseudarthrosis
- A first-degree relative (parent, sib, or offspring) with NF1 as defined by the above criteria
Common presentation of NF1 in Internal Medicine

- Malignancy: nerve sheath tumor, neurofibrosarcoma, soft tissue sarcoma, leukemia
- Neuropathy
- Stroke due to cerebrovascular stenosis
- Hypertension: renal artery stenosis and pheochromocytoma
A 32-year-old man, progressive proximal muscle weakness since the age of 7, wheelchair dependent at the age of 30, having two uncles affected by the same presentation and died at 50.

Q4: Investigation to obtain the right diagnosis?
A. Muscle biopsy for dystrophin staining
B. Creatinine kinase
C. NCV/EMG
D. Blood glucose, cholesterol and uric acid
E. PCR for SMN1 gene
Duchenne/Becker Muscular Dystrophy

- Onset: DMD at preschool age, BMD at 1st decade
- 1:3500 male birth
- Dystrophin gene on chromosome X
- X-linked recessive inheritance
- Symmetrical proximal muscle weakness with pseudohypertrophy of calves
- Early mortality by respiratory failure and cardiomyopathy
Duchenne muscular dystrophy
Myotonic Dystrophy type 1

- CTG repeat expansion on *DMPK* gene on chromosome 19
- Autosomal dominant
- ‘Myotonia’
- Frontal baldness, cataract and diabetes
- Cardiac conductive defect especially complete AV block
Facio-scapulo-humeral(-peroneal) Muscular Dystrophy

- Deletion within D4Z4 repeat region on chromosome 4
- Autosomal dominant inheritance
- Asymmetrical face, defects in blowing and whistling
- Uncommon for cardiac involvement
Limb-girdle Muscular Dystrophy

- Genetic heterogeneity
- Various clinical manifestation and onsets
- Proximal muscle atrophy of limbs and girdles
- Both autosomal dominant and recessive inheritance, rarely X-linked
- Mortality due to respiratory and cardiac failure
ทักทาย ถามชื่อ ถ้ามาเป็นคู่หรือครอบครัวให้ถามถึงความสัมพันธ์
เปิดประเด็นกับความคาดหวังในการมาตรวจครั้งนี้
เป็นโรคอะไร
เกิดจากอะไร
ความชุกเท่าไร/สถิติโรค
รักษาหายไหม
ถ่ายทอดอย่างไร
ความเสี่ยงเท่าไร
ปฏิบัติตัวอย่างไร (รวมถึง option การมีบุตร หรือเลี่ยงโรคในบุตร)
จะกลับมาดารงชีวิตเป็นปกติสุขในสังคมได้หรือไม่
เปิดโอกาสให้ซักถาม
ใช้ภาษาไทย ถ้าไม่มีคำแทนให้ขออนุญาตทับศัพท์