# Rare Disorders in Malaysia: Rare and Special

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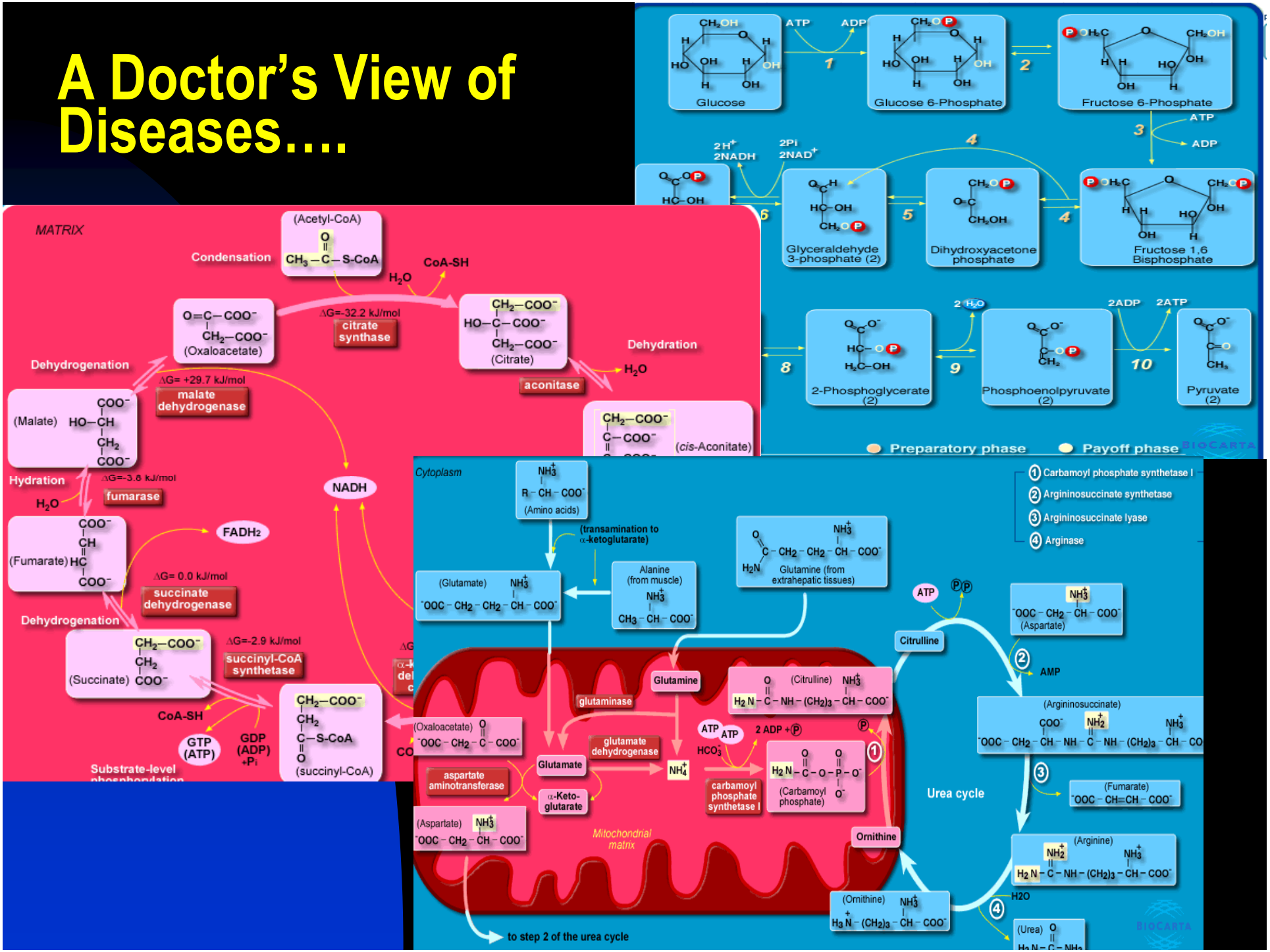
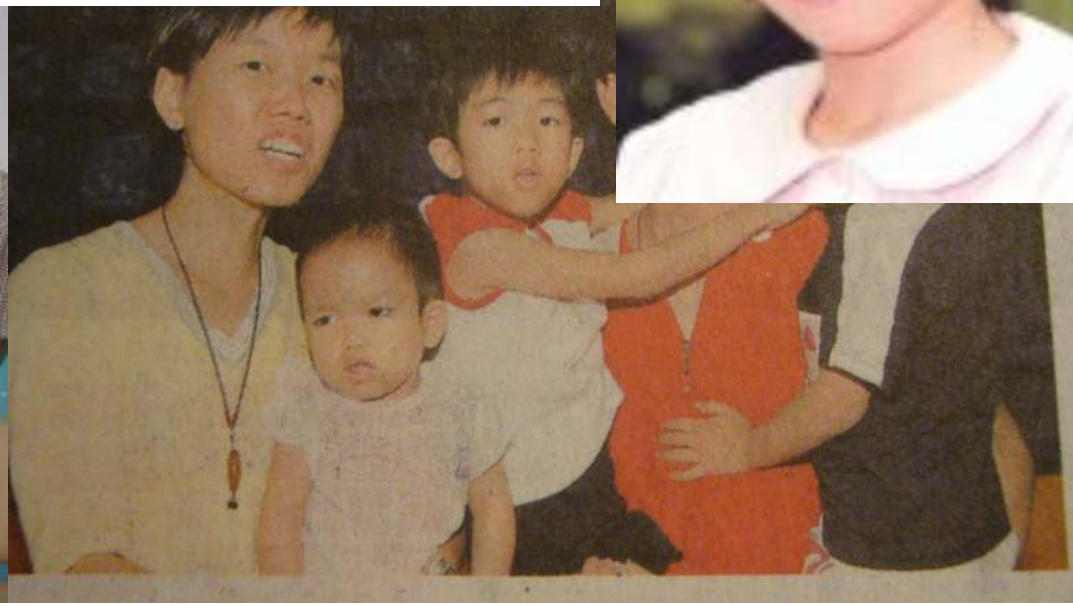
# A Doctor's View of Diseases….



# Rare Disorders


"My Children, My Family, My Community !"



# Rare Disorders: Myths & Facts

- About 3-5% of babies are born with serious birth defects. Each year, there are about 20,000 Malaysian babies born with birth defects
- Individually rare but collectively common
- Some of these can be recognised at birth, while some are diagnosed later in life
- Some are due to genetic changes, while others are due to both genetic and environmental factors
- These conditions cause medical and social problems and may recur again in some families

# Rare Disorders:

Problems:

Low scientific knowledge (false perception about rare disorders - ‘not important’ enough)

⇓

Absence of cure and treatment that could improve quality of life

⇓

Low knowledge amongst health professionals & public

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Negative social consequences

# What are Rare Disorders?

- Prevalence of less than 1 in 4000 people
- Examples:
  - O*steogenesis imperfecta* (brittle bone disease),
  - *Duchenne muscular dystrophy* (DMD),
  - *Prader-Willi* syndrome,
  - *Fragile-X* syndrome,
  - Mucopolysaccharidosis
  - Chromosomal abnormalities
  - Inborn errors of metabolism
  - Syndromes and many others.

# Many challenges…

- Limited educational resources on rare disorders for patients and family members especially in different languages
- Lack of public support groups for patients with rare disorders
- Limited professionals trained in early intervention programmes and services
- Lack of awareness and understanding among medical professionals, organisations and community on the needs of individuals with rare disorders

# More challenges…

- Limited funds to support treatment or to purchase special orphan drugs
- Limited genetic testing available locally
  - too costly to send overseas
  - insufficient laboratories and technologists
- Limited local research data on rare disorders

# Day to day challenges…

- Specific diagnosis
  - Rare unique syndromes – parents feeling alone in facing challenges
  - Breaks for parents – respite care for caregivers
- Coping with loss of child, grief and bereavement
- Dealing with feelings of guilt, shame and blame
- No specific diagnosis:
  - dealing with uncertainties
  - management based on needs
  - family planning issues

# Day to day challenges…

- Health maintenence
  - Routine paediatric care e.g. vaccination
  - Monitoring growth and nutrition
  - Ensure development  of skills
- Health surveillance
  - Hearing loss
  - Visual impairment
  - Difficulties in ambulating and getting around
  - Dental care
  - Personal hygiene and toilet

# More challenges…

- Sleep problems
- Behavioural challenges
  - Autism & Hyperkinetic disorder
  - Others: depression, aggression
  - Role of psychologist and psychiatrists
- Learning difficulties
  - IQ assessment: accurate?
  - Early intervention programme
  - Occupational therapy
  - Physiotherapy, speech therapy
  - Schooling?

# Jabatan Kebajikan Malaysia

- www.jkm.gov.my
- OKU registration
- Kemudahan pengangkutan awam
  - Tambang konsesi
  - MAS 50% penerbangan domestik
  - KTM 50% bagi semua kelas
  - LRT, KL Monorail 50%
  - Rapid KL – tiada bayaran
  - Transnasional 50%
- Pelepasan cukai sebanyak RM 5000 kepada pembayar cukai yang mempunyai anak OKU yang berusia dibawah 18 tahun
- Pelepasan cukai sehingga RM 5000 bagi membeli alat-alat khas untuk kegunaan sendiri,anak atau ibubapa OKU.
- Pengecualian Bayaran dokumen perjalanan

# Management of rare disorders

Gene ← Transplantation / gene therapy

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Protein ← Protein replacement

**Support for family & patient education**

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Symptoms ← Special diet; medications

**Support for family & patient education**

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Disease ← Medical or surgical treatment

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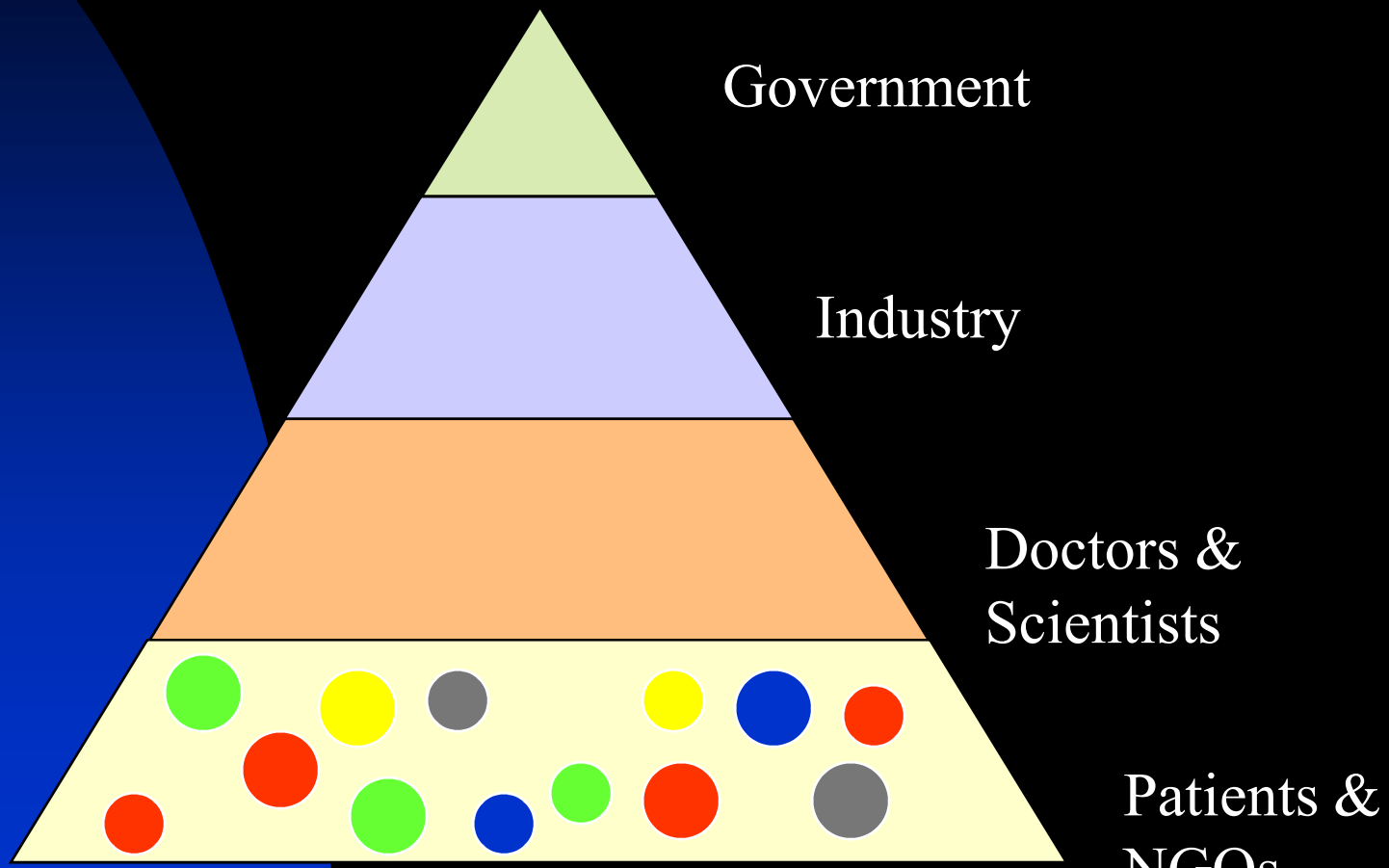
**Support for family & patient education**

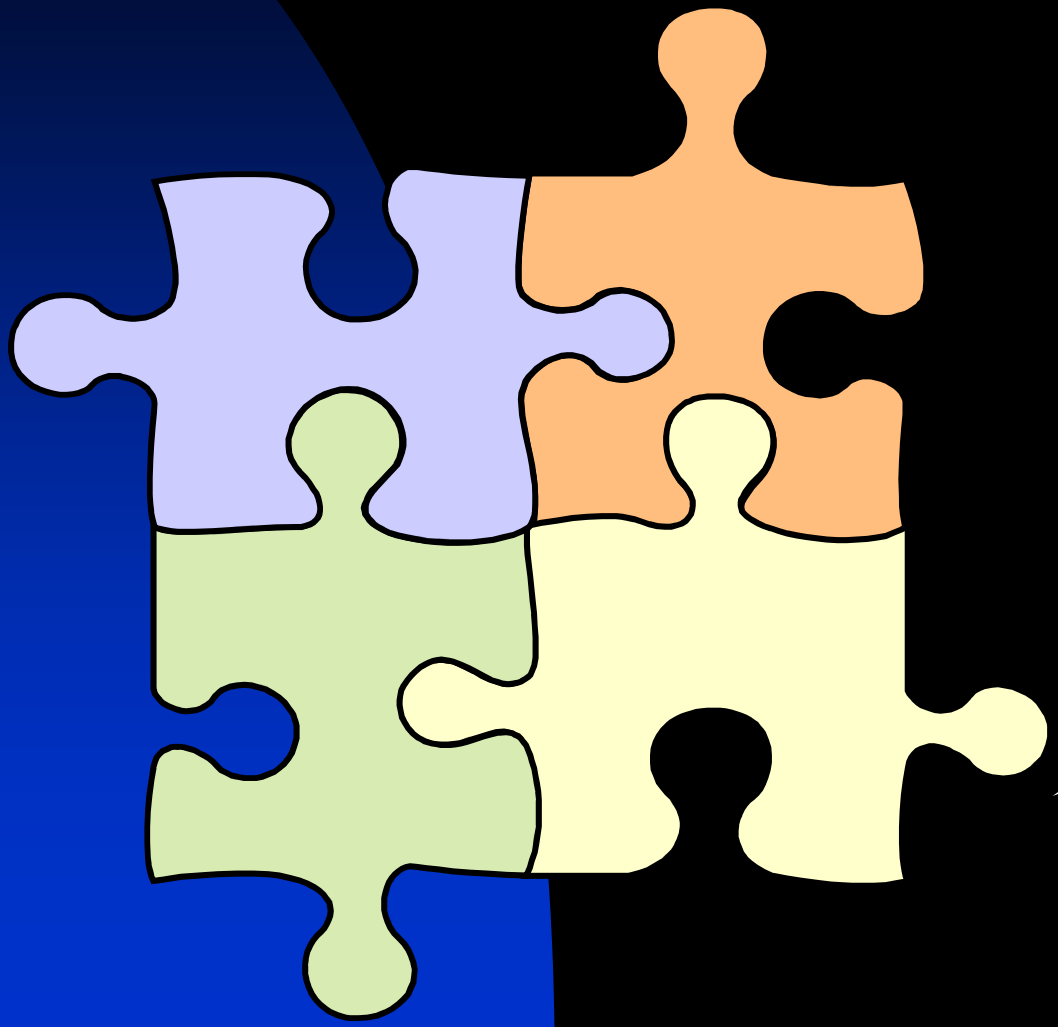
The family ← Genetic counselling; screening & genetic testing

**Support for family & patient education**

# Patients and their partners in health



# Rare Disorders:
# Putting the Jigsaw Puzzle Together



Patient Groups:

Equal partners

Share the burden

Speak with one voice

Work as a team

Network together

# MALAYSIAN RARE DISORDERS SOCIETY

(Registration No. : 0064-07-WKL)

# Our hopes…
# Our dreams…

Create a network among individuals and families with rare disorders.

Create a network among MRDS members with organisations, agencies and professionals involved in treating, educating and conducting research on rare disorders.

Serve as a contact and resource centre on rare disorders.

Promote the awareness on rare disorders among the Malaysian community.

Support and assist individuals and families affected with rare disorders in terms of welfare, treatment, rehabilitation, education and socials needs.

Collaborate with agencies and organisations that diagnose, research and treat rare disorders to increase the quality of life for the individuals and families affected.

Raise funds to support the activities and objectives of MRDS.

MRDS

# Thank you

# Our Team

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