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Clinical Features in Adult and Elderly Patients with ATR-X (Alpha-thalassemia X-linked Intellectual Disability) Syndrome: Report of Two Cases

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Introduction

ATR-X syndrome is characterized by severe developmental delay, alpha thalassemia, dysmorphic face, gastrointestinal problems, genital and skeletal abnormalities, result from mutations in the *ATRX* gene (Xq 13.3).

The prevalence is estimated 1 / 5800 to 7300 live born males and only about 300 patients reported worldwide.

Recognizable face of ATR-X syndrome leads to the genetic diagnosis, usually in childhood.

No longitudinal data suggests clinical course of aged ATR-X syndrome patients to date.

What this report adds

The distinctive facial appearance of ATR-X syndrome is preserved until adulthood. (Fig 1 and 2.)

There might be a lot of undiagnosed adult patients with recognizable face of ATR-X syndrome.

ATR-X syndrome related medial problems such as **feeding, gastrointestinal, urinary, cardiac and ophthalmological problems** also developed in adulhood. (Fig 3)

Lifelong interdisciplinary teams or comprehensive health care management should be considered to reduce mortality and morbidity in ATR-X syndrome.

Patient 1　61y male

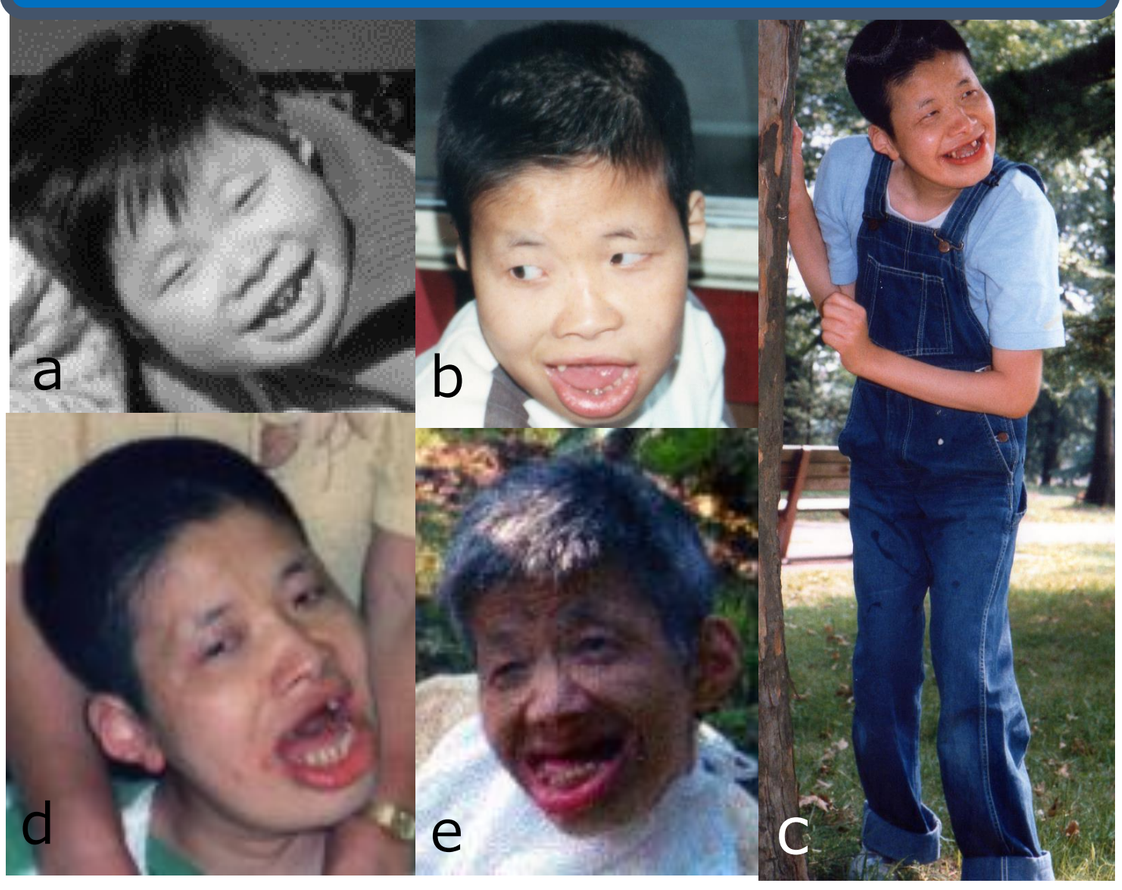

Fig 1.Photographs of patient 1.
(a)childhood, (b)　teen, (c) and (d) 20s, (e) 50s.

Informed consent had been taken from their family.

Patient 2　30y male

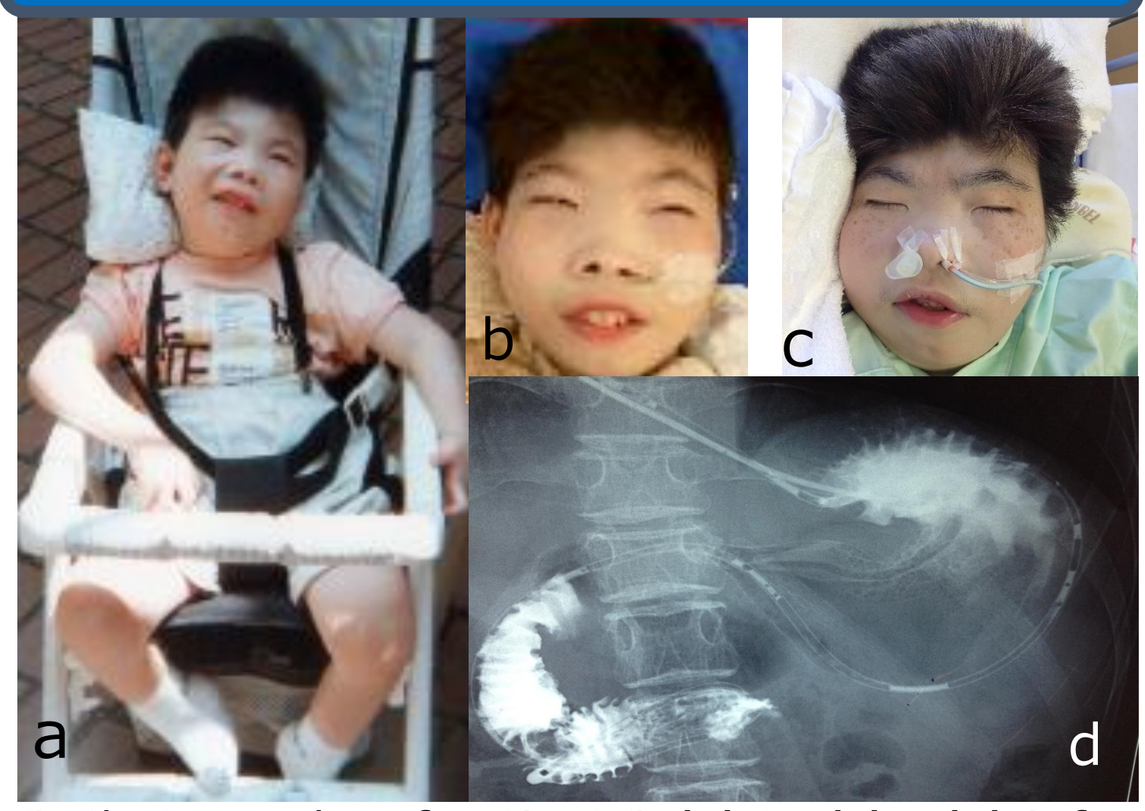

Fig 2.Photographs of patient 2 (a) to (c).　(a) infant, (b)　teen, (c) 30s. Abdominal X ray shows stomach lying near-horizontally suggests gastric rotation, and placed feeding tube in jejunum (d).

Fig 3. The age developed　clinical symptoms of ATR-X syndrome in our two patients. (**Red colored symptoms** developed at ≧ **18y**)　* y: year,　m: month

Clinical findings	Frequencies in ATRX syndrome(%)	Patient 1　(age*)	Patient 2 (age*)
Genetic diagnosis		c.832A>C mutation (54y)	+ (18y)
Psychomotor retardation	95	+	+
Intellectual skills		use few words and signs and aggressive behavior (childhood)	no word, smile and situational understanding (childhood)
History of motor development		**walk (20-43y)** **shaffling (43y)** **bedridden with respirator due to brain hypoxia (54y)**	sit with help (5y) bedridden due to repetitive pneumonia (10s)
Characteristic face	94	typical (childhood)	typical (at birth)
Mild Alpha-thalassaemia	90	+ no treatment	+ no treatment
Skeletal abnormality	90	drumstic phalanges, pes varus (childhood)	drumstic phalanges, talipes equinovarus, arthrogryposis (childhood)
Genital abnormality	80	none	micropenis, cryptorchidism (at birth)
Gastrointestinal	76	rumination (17y) **excessive dooling (27y)** **aspiration (47y)**	gastoesophagical reflux gastric pseudo-volvulus **recurrent ileus (24y)**
Feeding		**nasogastric tube feeding (54y)**	**nasogastric tube feeding (6m)** **nasojejunal tube feeding (23y)**
Seizures	35	transient (5y) no treatment	apnetic seizure (16y) oral therapy
Cardiac defects	20	septal defect **pulmonary hypertension (46y)** **pulmonary hypertension crisis　(54y)**	plumonary stenosis (at birth)
Urinary	15	bladder stone (13y)	**urinary toract infection (10-30s)** **bladder stone (19y)**
Others		**cataract (23y)**	airway obstruction (12y)

COI: none