Difference of Clinical Phenotypes and Immunological Features of 22q11.2 Deletion Syndrome in North-Eastern Thai Children Compare to Western Countries

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Background: *22q11.2 deletion syndrome is a common microdeletion syndrome that affected various systems.*
Objective: *To determine clinical phenotypes and immunological features of 22q11.2 deletion syndrome in north-eastern Thai children compare to western countries.*
Material and Method: *The authors described the clinical and immunological features in 20 north-eastern Thai children with 22q11.2 deletion syndrome that were followed-up at Srinagarind Hospital.*
Result: *Clinical phenotypes were facial dysmorphism (100%), congenital heart disease (80%) and cleft palate (30%). Prevalence of tetralogy of Fallot (TOF) in this syndrome was higher than in western. Serious infections were found including pneumonia, septicemia and brain abscess. Only a patient had panhypogammaglobulinemia and subsequently died. Selective IgA deficiency was not found. There was a twin patient conceived from intracytoplasmic sperm injection (ICSI).*
Conclusion: *TOF is more common in Asian patients than in western which different to selective IgA deficiency. The 22q11.2 deletion syndrome could be consequence from ICSI.*

Keywords: *22q11.2 deletion, Cleft palate, Immunoglobulin, Intracytoplasmic sperm injection, Tetralogy of Fallot, Hypothyroidism*

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A 22q11.2 deletion syndrome is one of the most common chromosomal microdeletion syndromes and the incidence is about 1 in 4,000 live births. The syndrome had been previously described as DiGeorge syndrome, velocardiofacial syndrome, conotruncal anomaly face syndrome and Cayler syndrome because of various phenotypes. However, the term 22q11.2 deletion syndrome is widely used instead of these in order to determine the cause of these syndromes. The syndrome is caused by a small deletion (about three million base pairs) of the q11.2 arm of chromosome 22 which can be detected by fluorescence in situ hybridization (FISH) technique. Most cases result from de novo deletion and the remainders receive the chromosome 22 with deletion at q11.2 arm from one of their parents. There is characteristic facial dysmorphism including narrowing of the height of the palpebral fissure, hypertelorism, widened nasal dorsum, hypoplastic nasal alae, short philtrum, micrognathia, malar flattening, malformed auricles, Pierre-Robin sequence, cleft palate and micrognathia[1]. Velopharyngeal insufficiency results in otitis media with effusion and delayed speech in children. Congenital heart defects were found in 22q11.2 deletion syndrome patients about 64-97% particularly conotruncal heart anomalies including tetralogy of Fallot (18-64%), ventricular septal defects (13-18%), interrupted aortic arch (4-16%), truncus arteriosus (2-10%) and transposition of great arteries (1%)[2]. Abnormal development of third and fourth pharyngeal pouches in this syndrome results in hypocalcemia from hypoparathyroidism and defect in immune system particularly cell-mediated immune response from aplasia or hypoplasia of thymus. Most of the patients have developmental delay in motor and language milestones. Immune system dysfunction in this syndrome usually presents as cell-mediated immune response defects caused by thymic hypoplasia or aplasia. T-cell

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lymphocytes are decreased by underdeveloped of thymus gland. However, immune function in most of the patients can be improved when they grow up. Less than 0.5% of the patients have severe immunodeficiency and need specific management[3]. Apart from the cell-mediated immune response dysfunction, there were previous studies that reported about abnormalities in humoral immune response in this syndrome including hypogammaglobulinemia, transient diminished IgG level, selective IgM deficiency and selective IgA deficiency[4-10]. Data from United States, United Kingdom and Italy show the prevalence of selective IgA deficiency in 22q11.2 deletion syndrome is about 2-13%[6-8,11].

The objective of this study is to describe clinical phenotypes and immunological features of 22q11.2 deletion syndrome in north-eastern Thai children and compare to previous studies in western countries.

Material and Method

The present study was descriptive study. The 22q11.2 deletion syndrome patients who were diagnosed by FISH technique with TUPLE1 probe and follow-up in genetic clinic during August 2012-July 2013 were invited to the study. All participants who join the study received the permission from their parents. Clinical history including birth weight, age at diagnosis, growth, development and history of infection, clinical phenotypes; congenital heart defects, Pierre-Robin sequence, cleft palate, and other organ anomalies were recorded and compared to previous studies. Laboratories tests results that include CBC, serum calcium, thyroid function tests were described. Immunoglobulin levels and CD3, CD4, CD8, CD19 lymphocytes sub-population were analyzed. Ethics committees at Khon Kaen University approved this study and informed consents were obtained from all participants and their parents.

Results

The age range of the patients was 0-16 years at the time of diagnosis with mean of 70.7 months (median of 57.5 months). Nine of twenty patients were male (45%). Nineteen patients (95%) were sporadic and only a female patient was inherited from an affected mother. One patient was born at 35 weeks of gestational age with dizygotic twins after intracytoplasmic sperm injection (ICSI) while another zygote has no deletion. Birth weight range was between 1.07-4.15 kg (mean of 2.63 kg and median of 2.77 kg).

Clinical phenotypes

All patients had subtle dysmorphic features. There were 6 patients with isolated cleft palate (30%), 2 patients with Pierre-Robin sequence (10%) and a patient with bilateral cleft lip and cleft palate (5%). Congenital heart defects were found in 16 patients (80%) Tetralogy of Fallot was found in 9 patients (45%). Three of them were tetralogy of Fallot with pulmonary atresia (15%). Other congenital heart defects were ventricular septal defect (20%), atrial septal defect (15%), truncus arteriosus (5%) and double outlet right ventricle (5%). Other congenital anomalies that found in this report were anorectal malformation, asymmetrical crying facies, indirect inguinal hernia, anterior glottic web and polydactyly. All congenital heart defects and other congenital anomalies were shown in Table 1.

Growth and development

Nineteen patients (95%) had height below 50th percentile and 12 patients (60%) below 3rd percentile. Two boys (10%) had obesity with BMI 31.39 and 31.61 kg/m^2 (above 97th percentile). Eleven patients were evaluated their developmental milestone and the result show that 8 patients (72.7%) had global delayed development. There were 3 patients with delay speech and language. IQ test was evaluated in 4 patients and it was found that only a patient had IQ 47 and the rest are in between average and borderline.

Laboratory results

CBC was evaluated in 18 patients and it was found that platelet counts were between 26,000 and 375,000/mm^3 (mean of 184,720/mm^3). Eight patients (44.4%) had thrombocytopenia (platelet count below 150,000/mm^3) while two of them were documented chronic ITP and received treatment with prednisolone. No patients had any history of severe hemorrhage. Thyroid function tests were evaluated in 13 patients and there were 4 patients (30.8%) had hypothyroidism and received thyroid hormone supplementation. Hypocalcemia (total serum calcium <8.5 mg/dl) was found in 12 of 19 patients (63.2%) with mean total serum calcium was 7.5 mg/dl while 8 patients (66.7%) need calcium replacement therapy. All laboratory results were shown in Table 2.

Immune status

Sixteen patients were evaluated for immuno-globulin levels. According to the evaluation, 8 patients had abnormal immunoglobulin levels as shown in Table 3. One patient had panhypogammaglobulinemia

Table 1. Clinical phenotype of north-eastern Thai patients with 22q11.2 deletion syndrome and comparison with other countries (%)

Clinical phenotype	This report	McDonald-McGinn et al 1999[20]	Repetto et al 2009[22]	Kitsiou-Tzeli et al 2004[23]	Oskarsdottir et al 2005[24]	Ryan et al 1997[25]	Tan et al 2008[26]
	Thai	US	Chile	Greece	Sweden	Europe	Singapore
Number of patients	20	250	208	17	100	558	17
Dysmorphic facies	100			82	100		
Congenital heart defects	80	74	59.6	65	64	75	94.1
TOF (all)	45	20	20.2	17.6	13	17	23.5
TOF with PA	15						
VSD	20	14	12	35.3	14	14	58.8
ASD	15	3.5	2.9		1	1	35.3
TrA	5	6	2.4		10	9	5.9
DORV	5				2		
Isolated cleft palate	30	11	13	11.8	6	9	11.8
Pierre-Robin sequence	10						
Laryngeal anomalies	5				1	1	
Asymmetrical crying	5						
Indirect inguinal hernia	5				13		6.3
Polydactyly	5				1	1	
Anorectal malformation	5				5	2	

TOF = tetralogy of fallot; PA = pulmonary atresia; VSD = ventricular septal defect; ASD = atrial septal defect; TrA = truncus arteriosus; DORV = double outlet of right ventricle

Table 2. Abnormal laboratory findings of 22q11.2 deletion syndrome patients

Abnormal laboratory results	% of patients
Thrombocytopenia (platelet <150,000/mm^3)	44.4
Idiopathic thrombocytopenic purpura	11.1
Hypothyroidism	30.8
Hypocalcemia (total serum calcium <8.5 mg/dl)	63.2

(patient No. 15) compared with normal range for age in Thai children[12]. However, selective IgA deficiency was not found in this report. Lymphocyte subpopulations were analyzed as percentage and cell-subset count on 12 patients compared with normal range for age in children[13] as illustrated in Table 2. Beside, 9 patients had abnormal lymphocytes subpopulation levels in percentage. According to cell-subset counts, only 4 patients had abnormal levels; patient No. 2 had increased CD8+ lymphocyte, No. 5 had decreased CD4+ and increased CD19+ lymphocyte, No. 16 had increased CD19+ lymphocyte, and No. 20 had decreased CD3+, CD4+ and CD19+ lymphocyte.

Infection

Fourteen patients (70%) had history of recurrent respiratory tract infection while 6 patients (30%) need admission for intravenous antibiotic due to bacterial pneumonia and one of them died from severe pneumonia and septicemia. Pathogenic organisms that could be isolated from 2 patients with bacterial pneumonia included *Klebsiella pneumoniae*, *Pseudomonas aeruginosa* and *Acinetobacter baumannii*. Chronic otitis media was found in 8 patients (40%). There was only a patient with brain abscess that was a serious infection apart from respiratory tract infection.

Discussion

This report shows clinical characteristic features of 22q11.2 deletion syndrome in Thai children with no sex preference. One patient is dizygotic twin and results of intracytoplasmic sperm injection (ICSI) while another neonate and her parents have normal phenotype too. She was born at 35 weeks of gestational age with birth weight 1,070 gram. She had bilateral cleft lip and palate, tetralogy of Fallot and anovaginal fistula. Even though there are previous reports about abnormal karyotyping after assisted reproductive technology especially ICSI entire pre and postnatal-diagnosis[14-17], the incidence of microdeletion syndrome follow assisted reproductive technology is still unclear. It was until recently only 2 reports in Pubmed database show the 22q11.2 deletion syndrome baby conceived through assisted reproductive technology[18,19]. Therefore, this report is showing that 22q11.2 deletion syndrome can occurs after ICSI. Further screening and studies about the incidence of 22q11.2 deletion syndrome and other microdeletion syndromes should be considered in assisted reproductive technology infants.

More than half of patients had short stature which their height below 3rd percentile. In this report, midline cleft palate was found in 30% of the patients that higher than other countries. McDonald-McGinn et al. reported cleft palate was found about 11% of the patients in United States[20]. Data in Chile showed cleft palate was 11.9-13%[21,22]. In Europe, cleft palate was range between 6-11.8%[23-25]. And in Asia, data from Singapore showed cleft palate was 11.8% of the patients[26]. The frequency of congenital heart defects in 22q11.2 deletion syndrome was not different from previous reports. However, the proportion of tetralogy of Fallot in this report (45%) is higher than in United States, Chile and Europe (18-30%) but lower than in Japan (64%) and Korea (54%)[2,20,22-25,27]. It appears that tetralogy of Fallot in 22q11.2 deletion syndrome in Asian is more frequent than Caucasion.

Other anomalies that found in this study were reported in the previous studies without significantly clinical difference. Because of nearly to one third of patients has global delayed development, so early intervention programs in patients with 22q11.2 deletion syndrome could be suggested. There are wide varieties of cognitive outcome in 22q11.2 deletion syndrome ranging from average to moderate mental retardation; but, the sample size of this study is too small and needs further studies in the future. Patients with hypothyroidism that need treatment are found in this report about 30%; as a result, we recommend that the thyroid function test should be performed in all cases of 22q11.2 deletion syndrome. According to autoimmune disease, only idiopathic thrombocytopenic purpura is found in 11.1% of patients which is more frequent than normal population.

Abnormalities of immunological profiles are found in 8 patients. One of them has panhypogammaglobulinemia and subsequently died before age one year old from severe bacterial pneumonia and septicemia due to *Klebsiella pneumoniae*, and *Acinetobacter*

Table 3. Peripheral blood lymphocytes subset and immunoglobulin level compare to normal range by age[12,13] and history of infections

Patients	Lymphocytes subset Percentage (absolute count)				Immunoglobulin levels				Recurrence, Chronic or serious infections	Organism
	CD3	CD4	CD8	CD19	IgG	IgM	IgA	IgE		
1	↔	↔	↔	↔	↔	↔	↔	↔	No	
2	↔	↓ (↔)	↑ (↑)	↔	↓ (slightly)	↑	↔	↔	Pneumonia, OME	
3	↔	↓ (↔)	↑ (↔)	↔	↔	↔	↔	↔	OME	
4	↓ (↔)	↓ (↔)	↔	↔	↑	↔	↑	↔	OME	
5	↓ (↔)	↓ (↓)	↔	↑ (↑)	↔	↔	↔	↔	No	
6	–	–	–	–	↑	↑	↔	↔	No	
7	–	–	–	–	–	–	–	–	Pneumonia, sinusitis	
8	–	–	–	–	↔	↑	↔	↔	No	
9	↓ (↔)	↓ (↔)	↔	↔	↔	↔	↑	↔	No	
10	↔	↔	↔	↔	↔	↔	↔	↔	Frequent URI	
11	↔	↔	↔	↔	↔	↔	↔	↔	Frequent URI	
12	–	–	–	–	↑	↑	↑	↔	Pneumonia, OME	
13	–	–	–	–	–	–	–	–	OME	
14	–	–	–	–	–	–	–	–	Pneumonia	
15	–	–	–	–	↓	↓	↓	↓	Pneumonia and septicemia (Death)	*Acinetobacter baumannii, Klebsiella pneumoniae*
16	↓ (↔)	↓ (↔)	↔	↑ (↑)	↔	↔	↔	↔	OME	
17	–	–	–	–	–	–	–	–	No	
18	↔	↔	↑(↔)	↔	↔	↔	↔	↔	Pneumonia, OME	*Pseudomonas aeruginosa, Klebsiella pneumoniae*
19	↓ (↔)	↓ (↔)	↔	↔	↔	↔	↔	↔	Frequent URI	
20	↓ (↓)	↓ (↓)	↔	↓ (↓)	↔	↔	↑	↔	Brain abscess, OME	

↔ = normal; ↓ = low; ↑ = high; OME = otitis media with effusion; URI = upper respiratory tract infection

baumannii. However there is not any information about lymphocyte subpopulation in this patient that be implicated about lymphocyte subpopulation, immunoglobulin level and infections. Other patients have diversely abnormal immunoglobulin levels including slightly decreased IgG and elevated IgG, IgM and IgA levels as found in previous study[28]. Selective IgA deficiency is the most frequent antibody deficiency in 22q11.2 deletion syndrome but it is noticable that selective IgA deficiency is not found in this study which is different from previous studies in western countries (United States, United Kingdom and Italy)[6-8,11]. According to 12 patients who are investigated both lymphocytes subpopulation and immunoglobulin levels, there is no clear correlation about lymphocytes subpopulation, immunoglobulin level and infection profile except patient No. 20. He had brain abscess and otitis media with effusion. Additionally, he had low level of CD3+, CD4+ and CD9+ lymphocytes and his immunoglobulin level showed only elevated IgA. However, he had underlying tetralogy of Fallot with pulmonary atresia which received palliative modified Blalock-Taussig shunt surgery since neonatal period and chronic otitis media that can increase risk for brain abscess too. However, limitation of this study is only 12 from 20 patients completely did both immunoglobulin levels and lymphocyte subpopulation studies. The reason is some patients lost follow-up before the date to investigate for their immunological profiles. So the actually prevalence of selective IgA deficiency and other abnormal immunodeficiency in north-eastern Thai children with 22q11.2 deletion syndrome may be need to study in the larger population.

There are 2 patients with serious infection. One has septicemia and severe pneumonia due to panhypogammaglobulinemia and the other has brain abscess as describe above. The most frequent infection in this study is chronic otitis media. Even though recurrent infection especially respiratory tract infection is frequent in 22q11.2 deletion syndrome, there is no correlation between site of infection and abnormal immunoglobulin levels in this study.

Conclusion and suggestion

This report shows clinical phenotype of Thai children with 22q11.2 deletion syndrome. The authors report 22q11.2 deletion syndrome in a baby conceived assisted reproductive technology; therefore, microdeletion syndrome should become aware and need furthermore studies about its incidence in children with assisted reproductive technology. Congenital heart diseases especially tetralogy of Fallot are more common than western. However, selective IgA deficiency is not as common as in western. The authors recommend an immunological evaluation including immunoglobulin levels in all patients. In addition, thyroid function tests should be performed in all patients. Chronic otitis media is the most common infection and also need early diagnosis and proper intervention. Regarding to recurrent and serious infection, both humoral and cell-mediated immunological studies are recommended in children affected with this syndrome and need furthermore studies.

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Potential conflicts of interest

None.

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ความแตกต่างของอาการทางคลินิกและลักษณะทางอิมมูโนวิทยาของผู้ป่วยเด็กกลุ่มอาการ 22q11.2 deletion ในภาคตะวันออกเฉียงเหนือของประเทศไทยเปรียบเทียบกับชาติตะวันตก

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ภูมิหลัง: กลุ่มอาการ 22q11.2 deletion เป็นความพิการแต่กำเนิดที่พบบ่อยมีความผิดปกติหลายระบบ

วัตถุประสงค์: เพื่อศึกษาลักษณะทางคลินิกและอิมมูโนวิทยาของผู้ป่วยเด็กกลุ่มอาการ 22q11.2 ในภาคตะวันออกเฉียงเหนือของประเทศไทยเปรียบเทียบกับประเทศทางตะวันตก

วัสดุและวิธีการ: การศึกษาเชิงพรรณาลักษณะทางคลินิกและอิมมูโนวิทยาของผู้ป่วยเด็กกลุ่มอาการ 22q11.2 deletion 20 รายที่ติดตามการรักษาที่โรงพยาบาลศรีนครินทร์

ผลการศึกษา: ผู้ป่วยทั้งหมดมีลักษณะใบหน้าที่จำเพาะ ร้อยละ 80 มีหัวใจพิการแต่กำเนิด และร้อยละ 30 มีเพดานโหว่ ความชุกของโรคหัวใจชนิด tetralogy of Fallot (TOF) ในกลุ่มอาการนี้พบมากกว่าในประเทศตะวันตก การติดเชื้อรุนแรงได้แก่ ปอดอักเสบ ติดเชื้อในกระแสเลือด และฝีในสมอง ผู้ป่วย 1 ราย มีอิมมูโนโกลบูลินต่ำทุกชนิดและเสียชีวิต แต่ไม่พบ selective IgA deficiency ในการศึกษานี้มีผู้ป่วย 1 ราย เป็นฝาแฝดและเกิดจากการทำ intracytoplasmic sperm injection (ICSI)

สรุป: TOF พบร่วมในผู้ป่วยเอเชียมากกว่าชาวตะวันตกซึ่งแตกต่างกับ selective IgA deficiency กลุ่มอาการ 22q11.2 deletion อาจพบได้ในเด็กที่ปฏิสนธิด้วยวิธี ICSI