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Original scientific paper

THE SPECTRUM OF MICRODELETIAN SYNDROMES AT THE HOSPITAL OF LITHUANIAN UNIVERSITY OF HEALTH SCIENCES

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Microdeletion syndrome is a rare condition which can be diagnosed by fluorescent *in situ* hybridization (FISH) method. We analyzed microdeletion syndromes cases during ten years period (2005-2015) at The Hospital of Lithuanian University of Health Sciences. We report 2 patients with Prader-Willi syndrome, 2 patients with Smith-Magenis syndrome, 1 patient with Angelman syndrome and 1 patient with Cri du Chat syndrome. All syndromes were confirmed by FISH. These cases contain mainly data about phenotype abnormalities and clinical symptoms.

Keywords: microdeletion syndrome, Fluorescence *in situ* hybridization (FISH), phenotype

INTRODUCTION

Microdeletion is a small chromosomal segment (<5Mb) deletion which involves loss of one or more genes. It causes multiple congenital abnormalities from phenotype alteration to mental retardation. Microdeletion syndromes are mostly sporadic due to novel mutations. Di George or Velocardiofacial (22q11.2), Angelman (15q11-13), Prader-Willi (15q11-13) and Williams syndrome (7q11.23)are the most common microdeletion syndromes. Also there are more known microdeletion syndromes such as Smith-Magenis (17p11.2), Cri du Chat (5p15.2) and Miller-Dieker (17p13.3) which are rare (HALDER *et al*., 2013; KURTOVIC-KOZARIC *et al*., 2016). Pathogenesis of microdeletian syndromes is different than monogenous diseases (SERAPINAS *et al*., 2012; SERAPINAS *et al*., 2013; DAUGELAITE *et al*., 2015).

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Because of less than 5 M deletion in chromosome, it is unable to be detected by conventional cytogenetic methods as karyotyping or traditional banding technique (PHILIP *et al*., 1994). In this case fluorescent *in situ* hybridization (FISH) is suitable for diagnosing microdeletion syndromes by using specific DNA probes.

Here we report six child individuals with microdeletion of six different unrelated families. There are two cases with Prader-Willi syndrome, two cases with Smith-Magenis syndrome, one case with Angelman syndrome and one case with Cri du Chat syndrome.

MATERIALS AND METHODS

A retrospective study was done in patients with microdeletion syndrome from the period of January 2005 to December 2015 in archives of Genetics and Molecular Medicine Department, at the Hospital of Lithuanian University of Health Sciences. All individuals belonged to Lithuanian families. The patients with microdeletion syndrome which was detected by FISH were selected for the study.

Genetic methodology description

FISH was performed using specific probes for the syndromes by standard protocols at Genetics and Molecular Medicine laboratory. All peripheral blood samples were received in heparin tubes and sample volume was 3 ml. The peripheral blood was cultivated using standard manufacturer's protocols (Euroclone, Milano, Italy).

One milliliter of blood was cultured in 5 ml of Chromosome Kit P medium (Euroclone, Milano, Italy) for 72 hours in 37°C incubator with 5% CO2. After harvest, FISH slides were dehydrated using different dilutions of ethanol and dried. A FISH probe was applied and incubated in Hybridizer (Dako, Colorado, USA), according to the manufacturer's instructions (Vysis, Abbott, Abbott Park, Illinois, USA). After the hybridization, coverslips were removed and the slides were washed with NP40 (0.03% solution) at 72°C for 2-3 min, followed by NP40 (0.01% solution) for 2 min at room temperature. DAPI was applied to the slides (4,6 diaminidino-2-phenylindole, Sigma, USA). For the FISH analysis, locus-specific probes of Prader-Willi/Angelman, Smith-Magenis and Cri du Chat were used. The cells were analyzed using a fluorescent microscope (Olympus BX61, Olympus, Tokyo, Japan) with Cytovision software (Cytovysion, AB Imaging, Germany). For each analyzed sample, at least 200 interphase nuclei were counted.

RESULTS

All patients had normal karyotype (boys 46,XY and girls 46,XX) except one patient with Smith-Magenis syndrome. Her karyotype was 46,XX,del(17)(p12). Differences and similarities of phenotype and clinical symptoms in patients with Prader-Willi, Angelman, Smith-Magenis and Cri du Chat syndromes are seen in Table 1.

Prader-Willi patient 1 (Patient 1)

The female patient was born at 39 weeks of gestation by caesarean section with a birth weight of 2964 g. She is a fist child in the family. After birth she was in incubator for two days because of poor thermoregulation. Since newborn period she had a poor sucking reflex and feeding difficulties. Also during neonatal period the patient had a weak cry, dry skin, a typical faces, hypotonia and hyporeflexia. Phenotype included almond shaped eyes, micrognathia, flat

face, and prominent nasal tip. Her developmental milestones were delayed. Moreover heart defect and small cysts in brain were monitored. For the patient Prader-Willi syndrome was diagnosed at 12 years age.

Her mother was 39 years old. During pregnancy the mother felt weak movements of fetus. In family history there are no genetic diseases.

Table 1. Clinical symptoms in patients with microdeletions

	Patient 1	Patient 2	Patient 3	Patient 4	Patient 5	Patient 6
Syndrome	Prader-Willi	Prader-Willi	Angelman	Smith-Magenis	Smith-Magenis	Cri du Chat
Dysmorphic features	+	+	+	+	+	+
Developmental delay	+	U/D*	+	U/D	+	+
Hypotonia	+	+	-	-	-	+
Cardiac findings	ASD**	ASD	-	ASD	-	ASD and VSD***
Cyst in brain	+	+	-	+	+	-

* Unknown data
** Atrial septal defect
*** Ventricular septal defect

Prader-Willi patient 2 (Patient 2)

The male patient was born at full term of gestation by caesarean section with a birth weight of 2956 g. He is a first child in the family. After birth the patient was weak and hypotonic. He had no suck reflex. Because of feeding difficulties newborn was fed by probe. During neonatal period he had hypoglycemia. Also he had a weak cry and poor reflex of spinal automatism. The facial features included flat and dysmorphic face, almond shaped eyes, prominent nasal tip and micrognathia. For the male newborn were monitored heart defect and small cysts in brain. At 1 month of age he was diagnosed Prader-Willi syndrome by FISH. The family history was not available.

Angelman patient (Patient 3)

The female patient was born at 37 weeks of gestation with a birth weight of 3250 g. In the time of birth she was in hypoxia. The girl is a first child in the family. During the pregnancy mother was diagnosed with anemia and preeclampsia. In postnatal period she was diagnosed with perinatal infection and jaundice. In addition, there was observed psychomotor retardation, feeding problems and sleep disorders. At age of 2 years she weighted of 12.3 kg (25^{th} centile) and her height was 87 cm (10^{th} centile). Short neck, wide chest and anomalous physique appear in phenotype of child. Additional she had sialorrhea and paroxysms of laughter. FISH analysis

showed a microdeletion of Angelman syndrome when she was 2 years old. The family history was noninformative.

Smith-Magenis patient 1 (Patient 4)

The female patient was born at 38 weeks of gestation with a birth weight of 2980 g. After birth she had stigma, jaundice and cranium defect. During postnatal period distinctive facial features as oblique eye slot, hypertelorism, ears were lower than eyes, irregular ear creeper, microgenia, hypoplastic lower part of the face and flat face were notable in the patient's phenotype. Further she had sandal gap deformity and partial cleft of second and third toes of both foot. A heart defect and brain small cysts were found in the newborn girl. Three months after birth Smith-Magenis syndrome was detected by FISH method. The family history is unknown.

Smith-Magenis patient 2 (Patient 5)

The female patient was born at full term of gestation with a birth weight of 3900 g. The girl was second child in the family. Older sibling was healthy. After birth there were no serious conditions. She was referred to the geneticist at 7 months for mixed developmental disorder and long-term cough. Phenotype particularity was flat face, hypertelorism, down-turned corners of mouth and high-arched palate. In addition, she had a forearm and elbow limitations and thicker hands. Global developmental delay was noticed at age of 7 months. The same age she was weighed of 9 kg (50^{th} centile) and her height was 69.5 cm (50^{th} centile). At age of 8 months she was diagnosed with Smith-Magenis syndrome using FISH method. The family history was significant for grandfather from mother's side with asthma.

Cri du Chat syndrome patient 1 (Patient 6)

The female patient was born at 37 weeks of gestation with birth weight of 1296 g. During the period of pregnancy assigned hypotrophy of fetus. After birth she was diagnosed with metabolic acidosis, muscular hypotonia and heart defects. In postnatal period she had frequent infections. Phenotypically she had microcephaly, trigonocephaly, lateral slot of eye were down-turned, mutual epicanthus, long filter, triangular slot of mouth, thin lips, and high and narrow palate, micrognathia, retrognathia and ears were lower than eyes. Moreover the girl had mutual clinodactyly of fifth finger, left palm crease, pes varus and narrow foot. At age of 4 years her height was 85.5 cm (<3th centile). In addition, her psychomotor development was delayed and she had sleep disorders. By FISH method Cri du Chat syndrome was detected when the patient was at age of 4 years old. The family history was not informative.

DISCUSSION

Before the year 2000 there were known only some dozens of microdeletion syndromes. Last two decades because of new technologies for high-resolution analyses more and more microdeletion syndromes were detected. Now it is known more than 200 microdeletion syndromes (WEISE *et al.*, 2012). According to data the most common microdeletion syndrome in humans is DiGeorge syndrome (22q11.2 deletion) which affects up 1 in 2000 live births (FUNG *et al.*, 2015).

Prader-Willi syndrome (PWS) has a prevalence of 1/15000 to 1/30000 (LANDAU *et al.*, 2016). The syndrome detection age is ranging from newborn to 15 years and even in adulthood

(ZHANG *et al.*, 2016). The major criteria list of Prader-Willi syndrome includes neonatal and infantile central hypotonia with poor suck reflex, feeding problems during infancy, excessive weight gain, dysmorphic face features which involves narrow face and almond shape eyes, hypogonadism, hyperphagia, global developmental delay and mental retardation. Also could be minor criteria: decreased fetal activity, behavior problems, speech articulation defects, sleep disturbance, thick viscous saliva and other individual indications (GUNAY-AYGUN *et al.*, 2001; BINGELIENE *et al.*, 2015). LANDAU *et al.* (2016) reported case about 3 weeks newborn with typical neonatal characteristics such as severe hypotonia, poor feeding, inability to suck, dysmorphic features and cryptorchidism. BIETH *et al.* (2015) described a patient with neonatal hypotonia, poor suck reflex and feeding difficulties, mild developmental delay, obesity, hypogonadism, aggressiveness and frequent temper tantrums. Our two presented patients (Patient 1 and Patient 2) with Prader-Willi syndrome had main neonatal symptoms as hypotonia, feeding difficulties with poor ability to suck and dysmorphic face with almond shaped eyes. There are evidences that early detection may contribute to some symptoms in early treatment with hormone replacement therapy (ANGULO *et al.*, 2015).

The frequency of Angelman syndrome (AS) is from 1 in 10000 to 1 in 20000 newborns (PANDA *et al.*, 2013). The age when Angelman syndrome was detected varies from 2 months to 12 years (BAI *et al.*, 2010; DUCA *et al.*, 2013; ZHANG *et al.*, 2016). The syndrome has three categories of consensus symptoms criteria: consistent features (100% of the patients), frequent features (>80% of the patients) and associated features (20-80% of the patients). Consistent features involve developmental delay, movement or balance disorder, behavioral uniqueness (e.g. frequent laughter/ smiling) and speech impairment. Other two categories includes delayed or disproportionate growth, seizures, abnormal EEG, flat occiput, wide mouth, suck/ swallowing disorders, feeding problems, abnormal sleep and other (DUCA *et al.*, 2013). BAI *et al.* (2010) announced about 17 patients of Angelman with developmental delay in movement, speech impairments and happy disposition. Some patients had seizures, abnormal EEG, microcephaly, wide mouth and flat occiput (BAI *et al.*, 2010). YIŞ *et al.* (2008) reported two cases about Angelman syndrome. Patients had motor and developmental delay, hypotonia, flat occiput, wide mouth, inverted nipples, strabismus, meaningless laughing, fever fluctuating between 37-39 ˚C and lots of infections since 2 to 6 months (YIŞ *et al.*, 2008). Our Angelman patient (Patient 3) had some similar symptoms as motor and development retardation, feeding problems, infections, slower growth and paroxysms of laughter.

Rate of Smith-Magenis syndrome (SMS) is from 1 in 15000 to 1 in 25000 births (GAMBA *et al.*, 2011). Smith-Magenis syndrome detection age varies from 6 months to 6 years (MONCLA *et al.*, 1991; GUPTA *et al.*, 2015) sometimes later (BLANCO-BARCA *et al.*, 2003; TSIRIKOS *et al.*, 2010; GAMBA *et al.*, 2011). By MONCLA *et al.* (1991) the symptoms of Smith-Magenis syndromes were described as mental retardation, behavioural problems, developmental delay (speech delay), facial dysmorphism, brachycephaly, a broad face with a flat midface, short and broad hands, hypotonia, hearing loss, subnormal EEG with slow rhythms. Behavioural problems may appear as hyperactivity, frequent temper tantrums, making abnormal noises with nose and tongue and insomnia (MONCLA *et al.*, 1991). From GUPTA *et al.* (2015) research could be added some supplement symptoms. In nine patients was seen clear facial phenotype including tented upper lip, broad forehead, midface hypoplasia, short philtrum and upslant of palpebral fissure. Also some patients had cardiac and renal malformations, cystic encephalomalacia, developmental delay and sleep disturbances (GUPTA *et al.*, 2015). Minor craniofacial anomalies,

self-injurious behavior, auto-amplexation (self-hugging), signs of peripheral neuropathy, dental anomalies, strabismus, deep hoarse voice, congenital heart disease and seizures also are added to clinical phenotype of syndrome (SCHLESINGER *et al.*, 2003; GAMBA *et al.*, 2011). Our Smith-Magenis patients (Patient 4 and Patient 5) had cranium defect, facial dysmorphism, flat face, thicker hands, heart defects, small cysts in brain and global development delay. Early diagnosis of Smith-Magenis and regular screening after diagnosis may prevent patients from severe scoliotic deformity (TSIRIKOS *et al.*, 2010).

Cri du Chat syndrome (CdCS) has a prevalence of 1 in 15000 to 1 in 50000 (AZMAN *et al.*, 2008). The diagnostic age ranges from 3 months to 6 years (DANGARE *et al.*, 2012; KHADER and HUNTLEY, 2013; KRGOVIC *et al.*, 2014). The clinical features contain low weight at birth, microcephaly, distinct facial dysmorphism, abnormal dermatoglyphics, hypotonia, feeding problems, scoliosis, flat foot, pes varus, cardiac and neurological abnormalities, syndactyly and typical high-pitched cat-like cry. Distinct facial dysmorphism includes round face, large nasal bridge, hypertelorism, epicanthal folds, downward slanting palpebral fissures, down-turned corners of the mouth, low-set ears and micrognathia. Metabolic anomalies (hyperglycinaemia) furthermore may occur. Moreover severe psychomotor and mental retardation, hyperactivity, self-injury, repetitive movements, hypersensitivity to sound, clumsiness and obsessive attachment to objects appear as behavioural disorders (MAINARDI, 2006). DANGARE *et al.* (2012) noted five patients with Cri du Chat who suffered from microcephaly, global developmental delay, facial anomalies (hypertelorism, mouth and palatal abnormalities and epicanthal folds), hypotonia, clinodactyly and congenital heart disease. Additionally they had behavioural difficulties: hyperactivity and social withdrawal (DANGARE *et al.*, 2012). Our Cri du Chat patient (Patient 6) had similar symptoms: low weight at birth, metabolic acidosis, muscular hypotonia, heart defects, microcephaly, facial abnormalities (down-turned lateral slot of eye, micrognathia and low-set ears), clinodactyly, pes varus and psychomotor developmental delay. Early identification of syndrome helps to adjust educational programs which improve many skills in childhood: ability to walk by them, make short sentences, feed and dress themselves (DANGARE *et al.*, 2012). Further an earlier diagnosis of CdCS gives possibility to cure orthopaedic anomalies at young age of patients (KHADER and HUNTLEY, 2013).

CONCLUSION

Microdeletions syndromes (Prader-Willi, Angelman, Smith-Magenis and Cri du Chat) appear in some cases of younglings in Lithuania. The majority of symptoms fit in general symptoms list but patients also have individual symptoms of the syndromes. Because of innovative technologies (FISH) syndromes are easier to identify. Other studies indicate that earlier detection of syndromes could help to prevent some complications during children developmental period and cure several defects. That shows an importance of diseases detection.

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SPEKTAR SINDROMA MIKRODELECIJA U "HOSPITAL OF LITHUANIAN UNIVERSITY OF HEALTH SCIENCES"

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Izvod

Sindrom microdelecije je redak slučaj koji može da se dijagnotikuje metodom flurescentne *in vitro* hibridizacije (FISH). Autori su analizirali slučajeve sindroma microdelecije u toku deset godina (2005 – 2015) u *The Hospital of Lithuanian University of Health Sciences.* U radu su prikazana dva (2) pacijenta sa Prader-Willi sindromom, dva pacijenta sa Smith-Magenis sindromom, jedan (1), pacijent sa Angelman sindromom i jedan (1) pacijent sa Cri du Chat syndromom i jedan pacijent sa svim sindromima koji su potvrđeni metodom FISH. Ovi slučajevi sadrže velikim delom podatke fenotipskim nenermalnostima i kliničkim simptomima

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