„Anthropological analyzes of ontogenesis in patients with lysosomal storage diseases, with particular consideration of mucopolysaccharidosis”

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2004 - **Licenciate**, Faculty of Biology, Adam Mickiewicz University in Poznan. Specializing in Human Biology dissertation title: Equipment of the grave and the biological status of the individual

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2013 – **PhD** degree of biological sciences in the field of biology – anthropology.
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PhD dissertation title: Pattern of growth characteristics of physical development in children with mucopolysaccharidosis type I and II (MPS I and MPS II)
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4. Scientific achievement submitted for the habilitation procedure.

4.1. Title of the scientific achievement submitted for the habilitation procedure

"*Anthropological analyzes of ontogenesis in patients with lysosomal storage diseases, with particular consideration of mucopolysaccharidosis*"

4.2. List of publications submitted for the habilitation procedure:

1. **Agnieszka Różdżyńska-Świątkowska;** Agnieszka Jurecka; Zbigniew Żuber; Anna Tylki-Szymańska. Can macrosomia or large for gestational age be predictive of mucopolysaccharidosis type I, II and VI? Pediatrics and Neonatology 2016 : Vol. 57, Nr 3, s. 181-187

Impact Factor ISI: 1.287

MNiSW: 20

My contribution is related to: anthropometric measurements, conception and design, conduct of the work, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 60%.

2. **Agnieszka Różdżyńska-Świątkowska**; Agnieszka Jurecka; Joachim Cieślik; Anna Tylki-Szymańska. Growth patterns in children with mucopolysaccharidosis I and II. World Journal of Pediatrics 2015 : Vol. 11, Nr 3, s. 226-231

Impact Factor ISI: 1.025

MNiSW: 20

My contribution is related to: anthropometric measurements, conception and design, conduct of the work, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 60%.

3. Zbigniew Żuber; **Agnieszka Różdżyńska-Świątkowska**; Agnieszka Jurecka; Anna Tylki-Szymańska The effect of recombinant human iduronate-2-sulfatase (idursulfase) on growth in young patients with mucopolysaccharidosis type II. PLoS One 2014 : Vol. 9, Nr 1, s. e85074

Impact Factor ISI: 3.234

MNiSW: 40.000

My contribution is related to: anthropometric measurements, conception and design, conduct of the work, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 30%.

4. Jezela-Stanek Aleksandra*, **Różdżyńska-Świątkowska Agnieszka***, Kulpanovich Anna, Ciara Elżbieta, Marucha Jolanta, Tylki-Szymańska Anna. Novel data on growth phenotype and causative genotypes in 29 patients with Morquio (Morquio-Brailsford) syndrome from central-eastern Europe. Journal of Applied Genetics accepted 7.03.2019

DOI: 10.1007/s13353-019-00491-1

*** equal contribution**

Impact Factor ISI: 1,756

MNiSW: 20,00

My contribution is related to: anthropometric measurements, conception and design, conduct of the work, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 30%.

6. Agnieszka Jurecka; Ekaterina Zakharova; Loreta Cimbalistiene; Nina Gusina; Vera Malinova; **Agnieszka Różdżyńska-Świątkowska**; Adam Gołda; Anna Kulpanovich; Gulnara Kaldenovna Abdilova; Elena Voskoboeva; Anna Tylki-Szymańska. Mucopolysaccharidosis type VI in Russia, Kazakhstan, and Central and Eastern Europe. Pediatrics International, 2014: Vol. 56, Nr 4, s. 520-525

Impact Factor ISI: 0.730

MNiSW: 15.000

My contribution is related to: anthropometric measurements, collective retrospective data, , interpretation of growth data, statistic analysis, making a graphs, drafting and revising part of article. I declare my contribution to be equal to 10%.

7. **Agnieszka Różdżyńska-Świątkowska**; Elżbieta Jurkiewicz; Anna Tylki-Szymańska Bioimpedance analysis as a method to evaluate the proportion of fatty and muscle tissues in progressive myopathy in pompe disease. JIMD Reports Seria: JIMD Reports, Volume 26

MNiSW: 5.0

My contribution is related to: anthropometric measurements, conception and design, conduct of the work, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 60%.

8. **Agnieszka Różdżyńska-Świątkowska** and Anna Tylki-Szymańska. The importance of anthropological methods in the diagnosis of rare diseases Journal of Pediatric Endocrinology and Metabolism. Received October 15, 2018; accepted January 29, 2019

DOI: https://doi.org/10.1515/jpem-2018-0433

Impact Factor ISI: 1,086

MNiSW: 15

My contribution is related to: anthropometric measurements, conception and design, conduct of the work, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 70%.

Total impact factor of publications which are scientific achievements submitted for the habilitation procedure: **8,941**

Total points of MNiSW for publications which are scientific achievements submitted for the habilitation procedure: **135**

4.3. Description of series publications which are the scientific achievement submitted for the habilitation procedure.

4.3.1. Introduction.

Lysosomal storage diseases

Lysosomal Storage Diseases (LSD) are group of diseases related with abnormalities in the functioning of lysosomes. Since the first lysosomal enzyme deficiency (Pompe disease, 1963) was indicated, more than 50 metabolic diseases in this group have been described and classified. They are caused by genetic defects causing a lack of severe deficiency in activity of enzymes required for the metabolism of lipids, glycoproteins or glycosaminoglycans. Due to their deviciency, undegraded or partially degraded macromolecules accumulates in lysosomes, causing dysfunction of cell, tissues and organs. Most LSDs are inherited in autosomal recessive manner, except of three diseases: Fabry disease, Hunter's disease (type II mucopolysacharyidosis) [1] which are X-linked recessive disorders, and Danon disease, which is an X-linked and dominant [2].

Occurrence of lysosomal storage diseases

Lysosomal storage diseases (LSD), due to their rarity of occurance, are called Rare Diseases or Ultra-rare Diseases, but taking into account the prevalence of LSD as a group of diseases, it is quite high and amounts to 1 in 7,000 - 8,000 live births (studies based on Australian and Dutch populations) [3, 4], in a study conducted in the Portuguese population, this frequency was 1 in 4,000 live births [5]. Due to the genetic determination of these diseases, a higher incidence of LSD was observed in specific ethnic groups, eg Gaucher disease and Tay-Sachs disease occur about 50-60 times more often in the Ashkenazic population than in the general population [6,7]. Mutations which cause a Rare Diseases is also characteristic for geographical regions, eg the Q70X mutation in the IDUA gene, underlying the type I of mucopolysaccharidosis, is much more frequent in Russia and Scandinavia than in Western Europe [8].

Lysosomal storage diseases show a wide spectrum of clinical symptoms. All lysosomal storage diseases are progressive. Most of them are characterized by symptoms from the nervous system with accompanying symptoms from other structures [9].

Pathomechanism and division of lysosomal storage diseases.

Lysosomal storage diseases due to the cause of the abnormalities can be divided into:

1. Caused by the abnormality in function of lysosomal enzymes - the basis is the lack or deficiency of enzyme activity leads to inhibition of degradation and in consequence, to the storage large macromolecules in lysosomes e.g. mucopolysaccharidoses type I, II, III, IV, VI, VII and IX, Pompe disease, Fabry disease, Gaucher disease.
2. Induced by defects of lysosomal transporters – defect of lysosomal membrane protein: e.g. Niemann-Pick disease, Lysosomal acid lipase deficiency.
3. Caused by abnormalities in the process of direct enzymes to lysosomes (defect of the proteins of endoplasmic reticulum membrane; Golgi apparatus or cytosol, which are involving in process of direct enzymes to the lysosomes) e.g. mucolipidosis type II, III and IV, ceroid lipofuscinosis)
4. Caused by the lack of activators of lysosomal enzymes - eg. Danon's disease [10].

Anthropology

The term anthropology means the science of the man and is derived from the Greek words anthropos (man) and logos (science). This is a broad concept that is currently used in two meanings: cultural anthropology, which is a science exploring culture, civilization, social systems and ethnic issues, and a physical anthropology concerning the biological properties of humans for example individual variety of body structure and activities. From the eighteenth and nineteenth centuries descriptions of the body, head and face individual features (anthroposcopy and anthropometry) were used in physical anthropology.

Anthropometry, the principal assessment technique in anthropology, makes it possible to collect and describe measurement data. Anthropometric techniques have been developed since ancient times. Even Hippocrates described various types of human heads and body structures. Gall's concept of phrenology contributed to the development of craniometry, while Charles White’s work is considered crucial in the development of somatometry. The first measurement instruments and indices were introduced by Paul Broca. He was the first to define the scope of anthropology and devise methods of anthropological study. The application of Francis Galton’s and Karl Pearson’s

statistical methods to biological sciences in general was a major breakthrough in anthropometry and gave rise to mathematical anthropology, developed in Poland by Jan Czekanowski. The year 1877 saw the publication of a list of the first 24 anthropometric pointslandmarks, whose names derived from Latin and Greek. Anthropometric measurement techniques were more precisely specified and generally standardized at the Third Congress of the International Anthropological Institute in 1927. The current measuring techniques have been systematized and described by Martin and Saller in "Lehrbuch der Anthropologie". Development of anthropology in Poland was particularly influenced by Jan Czekanowski. He used mathematical statistics it to calculate racial composition, compare different human populations, and infer their kinship, origin, migration directions, etc. A great contribution to the development of anthropological sciences was made by the creator of Polish auxology and co-creator of the global transdisciplinary human ecology, - Napoleon Wolański. He patented an instrument for measuring infants (liberometer) and developed two instruments for spatial spine measurements (kypholordosometer and spherodorsimeter) [11].

Anthropometry uses the assessment of certain morphological features to describe anatomical structure of the body. These features can be divided into qualitative, such as eye or hair color, and quantitative, such as body height and measurements of body parts. Qualitative features are assessed with the use of scales and templates, whereas quantitative features are measured with anthropometric instruments, the most important of which are: stadiometer, anthropometer, liberometer, various types of spreading calipers, sliding compass, skinfold calipers, anthropometric tape, and goniometers [11,12]. Anthropometric examination should be preceded by obtaining information on the subject's sex, date of birth, and a history of his or her physical development. Moreover, the information on the place of birth of both the subject and his or her parents may be useful. Anthropometric examination involves measuring the distance between certain landmarks on the human body. These anatomical landmarks are the most important element of anthropometry, and knowing their location is crucial for understanding the descriptions of individual measurements and the way they are taken [12].

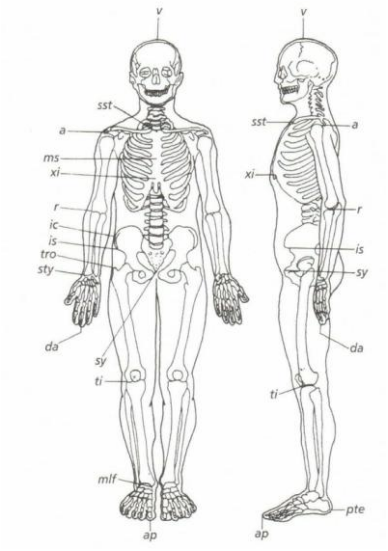

Fig. 1 The most important measurement points on human body (Malinowski, Bożiłow, Fundamentals of Anthropometry, Methods, Techniques, Standards, Scientific Publishers PWN, Warsaw-Łódź 1997)

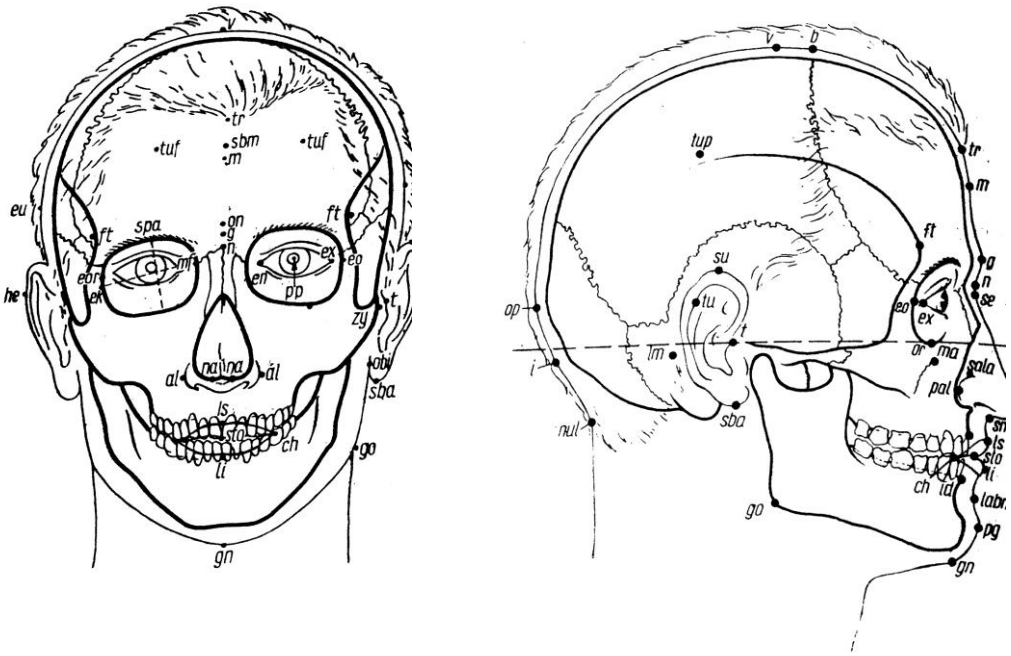

Fig.2. Anthropometric points of the head (Malinowski, Bożiłow, Fundamentals of Anthropometry, Methods, Techniques, Standards, Scientific Publishers PWN, Warsaw-Łódź 1997)

Anthropological methods in biology and medical biology

In biological studies, it is extremely important not only to describe the features of "normal" organisms (individuals with values of observed characteristics from the norm range), but also organisms with features different from the averages. Only then, is it possible to understand the

biological regulating processes and mechanisms. In case of the majority of biological system studied, understanding the function of an organ, tissue or gene is possible through research in which predetermined modifications are introduced, for example mutations in specific genes. For obvious ethical reasons, such experiments are not possible in the case of human research. That is why information about the functions of human genes comes mainly from animal studies or from studies of naturally occurring mutations in the human population. These mutations, often cause specific genetic diseases, so they are interesting for both biologists and representatives of medical sciences.

In order to assess a patient's physical development, it is necessary not only to describe his or her body structure, but also to interpret these findings, taking into account reference standards for the prevalence of particular features in the population. There are several methods of assessing an individual's development on the basis of current standards. These methods involve the use of tables of arithmetic means of selected parameters for a given age (a method introduced by Clemens von Pirquet) table method, or graphic methods of estimating the rate and harmony of development , or morphogram method [12]. However, the most popular and most common reference standards for physical development are in the form of growth charts. WHO recommends international height and weight charts; however, many researchers suggest that the use of international charts might not be suitable in some cases and may lead to underestimating the number of people with height deficit in the population [13–15]. Apart from height and weight anthropometric measurements include also other parameters. Describing a subject's phenotype or physical development, anthropologists focus primarily on proportions in the body's structure as well as any deviations from reference standards. One very useful means of representing inter-individual variations in body structure in the case of rare diseases are indices, which are ratios of two measurements. Such indices express the range of a given variation.

The importance of anthropological analyzes in the description of the biological state in genetic diseases

From the point of view of embryogenesis, the face and distal limb segments are among the most structurally complex parts of the body. There is a physical interaction between cerebral and facial development, therefore a number of central nervous system anomalies are directly related to certain dysmorphic facial features; such as in the case of holoprosencephaly and orbital hypotelorism [16]. Nearly half of all children with intellectual disabilities exhibit at least some body structure (mainly facial) anomalies, most of which are classified as minor physical anomalies. It is not always the case that facial features or body structure are characteristic enough to immediately arouse suspicion of a disease. In some cases they may be less obvious, particularly in the early

postnatal period; moreover, in milder forms of the disease, its phenotypic features can be very discreet. In spite of the undeniable advantages of modern diagnostic techniques, which help accurately and reliably verify the genetic background of a disease, it is the clinician who plays the primary and fundamental role in the diagnosis of these diseases. The difficulty in assessing dysmorphic features causes many rare diseases to remain undiagnosed or be diagnosed too late. Despite the indisputable benefits and accuracy of modern diagnostic techniques (e.g. molecular genetic testing)availability of diagnostic tools in the form of lists of co-occurring features specific to particular syndromes or computer-based diagnostic programs, genetic syndromes that involve of facial dysmorphism are usually first diagnosed by a clinician on the basis of a characteristic facial appearance. However, this is a subjective assessment, which depends on the skills and experience of a given clinician. For these reasons, objective methods of assessing facial features and body structure are extremely desirable in making accurate clinical diagnoses; hence the invaluable role of anthropology in medicine. In addition to helping establish the diagnosis, anthropology makes it possible to describe the natural history of the disease more accurately. Our understanding of the natural history of the disease (i.e. the consecutive pathological changes over the course of the disease) helps us evaluate physical development at individual phases of ontogenesis, describe the structure and proportions of the body, and characterize dysmorphic features and other changes which take place over the course of the disease. A majority of rare diseases are caused by a genetic metabolic defect, which can have different effects on different cells and tissues. This disrupts the harmonious development of an individual viewed as an integrated system. A given metabolic disease may be expressed to varying degrees in different organs. For instance, some metabolic diseases produce more pathological changes in bones than in the liver, in other cases muscles are affected more than the central nervous system. Disease-induced changes in body morphology, including variations in height, weight, and proportions, become the markers which allow an anthropologist to identify abnormalities and objectively assess the magnitude and direction of deviation from the normal developmental pattern. Therefore, knowing the natural history of a given disease, we could establish a model for how a particular disease develops, which would allow us to assess and better understand the occurring changes. This model would become a reference tool to help assess the development of affected individuals and predict its subsequent stages. Identifying developmental disorders at individual phases of ontogenesis, describing the anomalies, and adapting them to create such a model have to be carried out accurately and objectively; this requires suitable tools and knowledge.

4.3.2. Description of series publications which are the scientific achievement submitted for the habilitation procedure

Publication no 1 (Can macrosomia or large for gestational age be predictive of mucopolysaccharidosis type I, II and VI?) The first publication from the series assessing the birth parameters of children with mucopolysaccharidosis and their comparison with a healthy population. Mucopolysaccharidoses (MPS) are a group of lysosomal storage disorders caused by a deficient activity of enzymes responsible for the catabolism of glycosaminoglycans (GAGs). Partially degenerated GAGs are accumulated in lysosomes, causing dysfunction off cells, tissues and organs [1]. Mucopolysaccharidosis are progressive diseases, characterized by many clinical symptoms, although they may differ in severity various types. Clinical symptoms include a series of changes in the skeletal system called *dysostosis multiplex* caused by abnormalities in osteogenesis. Characteristic are also: enlargement of the liver, spleen, tongue, thickened and prominent facial features. In the course of the disease hearing and sight impairment occurs (corneal opacity, optic nerve damage), as well as abnormalities in the respiratory and cardiovascular systems (defects of the heart valves, cardiomyopathy, cardiomegaly), limitation of joint mobility, associated with, among others, progressive contractures. Mental retardation is observed in MPS I Hurler, MPS II (severe form), all subtypes MPS III and MPS VII [1, 17-19]. The occurrence of mucopolysaccharidosis in the population is difficult assess, from available sources the occurrence is estimate at 1 in 22,500 live births [20].

In **first publication** we compare mean values for birth body length and weight between patients with MPS I, II, VI, and the general population and this parameters were greater than in the general population. The pathomechanism of this phenomenon is still unclear. In patients with MPS I and II, accumulating GAGs are heparan sulfate (HS) and dermatan sulfate (DS). One hypothesis speculates that the over-growth in fetal and early postnatal life could be connected to the fact that HS, acting as a coreceptor, binds to several proteins, including growth factors. An increased level of HS might therefore overstimulate axial bone growth in children with MPS at early developmental stages [21]. Children with MPS II, whose HS is one of the accumulated GAGs, had usually a higher or normal stature for their age [22-24]. On the other hand, the accumulation of DS over time would cause an inhibition analysis of MPS growth plates showing clusters of enlarged, GAG-containing cells that disrupt a normal columnar architecture of growth plate cartilage, presumably leading, in part, to abnormal bone growth. Simonaro et al suggested that the main tissue of these disorders is the cartilage rather than bone itself [25]. Hinek and Wilson, however, reported that the process of elastogenesis takes place in the shaft of long bones during fetal life and accumulations of DS lead to early disruption of normal elastogenesis [26]. **In publication no 1 the birth data of patients with mucopolysaccharidosis were analyzed for the first time.** At the time of birth, many MPS patients are larger than the general population. High birth length can be suggestive of MPS disease and should

raise suspicion aiding in early disease recognition. The age of diagnosis is usually between 9 months and 36 months, but may be significantly delayed in attenuated forms. Including MPS disease in the diagnostic algorithm of LGA and macrosomia may increase the chances of diagnosing these patients very early in life, which is crucial for efficient treatment. Despite the fact that children with MPS are born large, the rate of growth decreases in the first year of life, and one of the symptoms of the disease is short stature. Human growth is a multi-factorial and complex process, involving physiological interplay between nutritional, endocrine, and metabolic factors, on a wider background of variation in genetic traits and environmental exposure. MPS diseases lead to a profound disruption in normal mechanisms of growth and development. The underlying cause of degenerative bone and joint disease is a lack of skeletal remodeling, disordered endochondral and intramembranous ossification, disruption of normal elastogenesis, and the infiltration by GAGs of the ligaments, tendons, joint capsules, and other tissue structures. GAG storage in MPS induces a complex sequence of molecular abnormalities leading to inflammation, apoptosis (cartilage), and hyperplasia (synovial membranes), resulting in poorly organized and metabolically abnormal connective tissue matrices. Currently, enzyme replacement therapy (ERT) has become available for patients with MPS I, II, IV and VI and knowledge about the natural history of MPS is necessary for a reliable evaluation of the effects of its treatment.

Study nr 2 (**Growth patterns in children with mucopolysaccharidosis I and II) This is the first published publication in the world which objectively evaluating the development of children with MPS I and II.** And showing the pattern of their development which significantly differs from the pattern of healthy children growing. Mucopolysaccharidosis I (MPS I) is caused by a deficiency of alpha-L-iduronidase (IDUA; EC 3.2.1.76) lysosomal glycosidase involved in the degradation of heparan sulphate and dermatan sulphate [1]. Depending on the severity of MPS I clinical phenotypes ranges from the most severe, which is Hurler syndrome, to the attenuated, Scheie syndrome. In all variants there are significant differences in the degree of central nervous system involvement. It is a progressive disorder with multiple organ and tissue involvement that leads to death in childhood. An infant with Hurler syndrome appears normal at birth but may have inguinal or umbilical hernias. Diagnosis of Hurler syndrome is commonly made between 4 and 18 months of age; a combination of skeletal deformities, recurrent ear and nose infections, inguinal and umbilical hernias, coarse facial features, hepatosplenomegaly, and enlarged tongue first. This mild form of MPS I is characterized by joint stiffness, aortic valve disease, corneal clouding, and few other somatic features. The facial features are characteristically coarse, but intelligence is normal. The joint involvement is prominent in the hands with a claw-hand deformity. MPS I Scheie patients can have a stiff painful foot, pes cavus, and genu valgum [1]. The gene for α-L-iduronidase, called IDUA, is located on chromosome 4,

in position p16.3. It is composed of 14 exons and has a length of about 19,000 base pairs. The two most common mutant alleles are W402X and Q70X, as well as the less frequent allele, P533R. Their product is an abnormal enzyme, and their occurrence in a homozygous or heterozygous form is responsible for severe form of MPS I [27, 28]. Mucopolysaccharidosis type II (MPS II, Hunter disease, OMIM 309900) is an X-linked recessive disorder caused by mutations, resulting in a deficient activity of the lysosomal enzyme iduronate-sulphatase (IDS, EC 3.1.6.13). IDS is one of the enzymes involved in degradation of the glycosaminoglycans (GAGs) dermatan sulphate (DS) and heparan sulphate (HS). Impaired GAG degradation results in the lysosomal accumulation of DS and HS leading to progressive damage of various tissues and organs [29]. Affected individuals present with coarse facial features, short stature, skeletal deformities, joint stiffness and frequently mental retardation. Attenuated and severe clinical variants differ in the age at onset, presence or absence of progressive intellectual deterioration and survival time [1, 18]. Patients usually become symptomatic at the age of 2–4 years. MPS II in women is very rare and is secondary to the presence of either pathogenic mutations in both alleles of the IDS gene or nonrandom inactivation of the X chromosome in heterozygotes [20]. MPS II is inherited as an X linked recessive and the IDS gene, coding for idursulfase, is located at position q28 [30, 31]. The results **of publication no 2**: **Growth patterns in children with mucopolysaccharidosis I and II** showed the existence of a characteristic downward trend in a degree and direction of deviations in MPS boys when compared with the Polish reference charts. The mean z-score values for every MPS group showed that until the 24th month of life, the growth pattern for all MPS I and II patients was similar and the average z-score values for body height were greater than the reference charts. Afterwards, growth patterns began to be different for individual groups. For boys with Hurler syndrome, the body height below the 3rd percentile was reached after the 24th month of life, for patients with severe MPS II between the 6th and 7th year of life, and for patients with attenuated MPS II between the 8th and 9th year. After this initial period of intensive growth, body height in subsequent years for patients with MPS I and II both reached significantly lower values when compared with the reference charts. Although this trend has been corroborated by earlier publications, there is still a shortage of complementary investigations that analyze the physical development of children with MPS and carried out by appropriate specialists. The reasons for differences in growth dynamics between patients with MPS I and II during first years of life remain unknown. A greater amount of toxic DS deposited in the cartilage in patients with MPS I could explain this process. The growth patterns in this study present a reference model of growth. In this way, it has been proven that the severity of the disease can be assessed based on the dynamics of growth. In patients with a severe form of the disease, growth stopped faster as compared to patients

with atenuated form. Knowledge of the natural pattern of growth of patients with mucopolysaccharidosis allows to objectively assess the effect of treatment on body height.

In publication no 3 (The effect of recombinant human iduronate-2-sulfatase (idursulfase) on growth in young patients with mucopolysaccharidosis type II) the study objective was to evaluate the effectiveness of idursulfase on body height in young patients with MPS II by comparing a natural growth pattern in children with MPS II naive to ERT with growth of MPS II patients who started ERT before 6 years of age. The growth data from 13 patients treatment before the age of 6 was compared with retrospective data from 50 people who were never treated enzyme replacement therapy. This is an important work because the introduction of enzyme replacement therapy for mucopolysaccharidosis type II, assumes the improvement of many parameters, including body height. Process of growth is non- linear with both periodic and chaotic elements, which are specific for growth pattern in human species. Velocity and rate of growth change during ontogeny and growth rate differs in particular age classes. Generally, growth can be characterized as a period of accelerated growth divided by periods of slow growth. Individuals grow according to genetically determined pattern. The growth pattern contains the following stages: infancy with rapid postnatal spurt immediately after birth; childhood followed by a small mid-growth spurt at the age of 6–7 years; juvenile and adolescence with its marked pubertal growth spurt. Patients with MPS II, despite the disease, show the same general growth pattern, characteristic for the human species. In connection with the above, apart from showing the growth curves on the percentile charts for each patient, the analysis of the dynamic of growth was carried out. To show the degree and direction of deviations of studied features in children with MPS II, the data for group 1 and 2 was divided into 24 calendar age classes and standardized for polish reference charts. **No apparent difference was observed between the growth rates in treated patients and the untreated disease. New approaches for the management and treatment of MPS disorders are necessary to influence skeletal abnormalities.**

Study no 4 (**Novel data on growth phenotype and causative genotypes in 29 patients with Morquio (Morquio-Brailsford) syndrome from central-eastern Europe**. Based on presented herein clinical re-evaluation of 29 patients with MPSIVA, with focus on anthropometric and molecular data, we like to underline the importance of detailed anthropometric assessment, which add important, objective clinical details, being valuable even for prediction of the phenotype. We do believe that such analyses, if performed as a rule in others rare inherited conditions as well, give novel insight into the phenotypic characteristics, and natural history of any of them. Mucopolysaccharidosis type IV A (Morquio syndrome A, OMIM MPS IVA 253000) is an autosomal recessive disorder caused by deficient activity of the lysosomal enzyme galactosamine-6-sulfatase (GALNS; 612222). In the

absence of enzyme activity, stepwise degradation of keratan sulfate and chondroitin-6-sulfate proceeds, resulting in intracellular accumulation of these glycosaminoglycans (GAGs) into the lysosomes, leading to progressive disorder with multiple tissue and organ involvement. The predominant clinical features of Morquio syndrome are those related to the skeleton and their effects on the central nervous system. As in most mucopolysaccharidoses, patients with Morquio syndrome appear normal at birth. Typical skeletal anomalies of Morquio syndrome include dwarfism with short trunk; platyspondylia; odontoid hypoplasia; kyphosis; hyperlordosis; scoliosis; ovoid deformities of the vertebrae; genu valgum; ulnar deviation of the wrist; valgus deformity of the elbow; inclinations of distal ends of radius and ulna toward each other; deformities of metacarpals and short phalanges; epiphyseal deformities of the tubular bones; widened metaphyses; and osteoporosis. Joints tend to be hypermobile secondary to ligamentous laxity, but decreased joint mobility can occur in the large joints. The GALNS gene, coding for 6-N-acetylgalactosamine sulphate sulfatase, has a length of over 40,000 base pairs and consists of 14 exons, and MPS IVA lies in many different point mutations or small deletions [1, 32]. Following birth, children with MPS IVA grow slowly, and reach their final height at approximately 8 years, the final height of the body rarely exceeds 100 cm [33]. The differences in body proportion between healthy and affected children increased with age. In our study **(no 4)** following conclusions could be drawn. Body length at birth was statistically greater in children with this disorder than in the general population. Body height below the 3rd percentile was reached earlier by boys than girls (after 24th month vs. 4th and 5th year of life), during first 5 years of life females lost about 10 cm while boys in this same period - about 18 cm. Dynamics of head circumference grow was grater in a group of girls than in boys, with mean head circumference in adulthood grater among girls. Although anthropometric deficiencies increased with age of MPSIVA patients, the height within normal range is however possible, depending on the causative *GALNS* variant. Regarding the molecular data, apart from novel pathogenic variants identified in MPSIVA patients from Poland and Belarus (listed below), genotype-phenotype analyses showed that: milder (attenuated) forms of mucopolysaccharidosis type IVA might result from substitution c.280C>G (p.775C>G) and c.680delT (p.Phe227Ser*92), the less severe growth deficiencies were supposed to result from c.280C>G (p.775C>G) and may be observed in heterozygotes c.121-9T>G(;)680delT. Variant c.280C>G (p.Arg94Gly) influences more severe MPSIVA expression. The most frequent variants in our central and eastern European population were c.502G>A (20%) and c.1156C>T (17%), moreover 5 novel molecular variant in GALNS were identifies - c.121-9T>G (p?), c.547G>T (p.Asp183Tyr), c.680del (p.Phe227Ser*92), c.1427A>C (p.Gln476Pro), and c.1482+5G>A (p?).

In patients with mucopolysaccharidosis type VI (Maroteaux-Lamy Syndrome), caused by a deficiency of arylsulfatase B (ARSB; EC 3.1.6.12) involved in the degradation of dermatan sulfate, the central nervous system is also not affected by the disease. The rate of clinical progression of MPS VI varies considerably, generating a wide continuous clinical spectrum from severe to relatively attenuated. The most prominent and debilitating manifestations usually arise in the musculoskeletal, cardiorespiratory and nervous systems, but individual cases vary considerably in the timing of symptom onset, the pattern of organ involvement, and the rate of disease progression [1, 34]. During the last 30 years, 600 patientw have been recognized in the world, 7 in Poland. The body stature characteristic for this type of MPS has been described in several publications in which the dr Rozdzynska-Swiatkowska was a co-author [35-37]. To the series of publications has been included the unique study, because it contains analyzes of a huge group of patients for such a rare disease, **publication no 5 (Mucopolysaccharidosis type VI in Russia, Kazakhstan, and Central and Eastern Europe.).** The study objectives were: to describe the natural history of MPS VI (n = 49) with regard to clinical manifestations of the disease, growth patterns, and relationship between height, glycosaminoglycan (GAG) level and severity of the disease; to analyze the ARSB mutations in MPS VI patients from Russia (n = 13), Kazakhstan (n = 2), Czech Republic (n = 2) and Poland (n = 1); to assess the frequency of the p.R152W mutation among the Russian patients (n = 17) and in the whole series (n = 49); (iv) to assess the MPS VI prevalence and incidence rates in Poland, Lithuania, Belarus and Estonia; and (v) to assess the carrier frequency of p.R152W mutation in the ARSB gene in the Polish and Lithuanian population. Patients included in this study had the same general pattern of disease progression as observed in MPS VI patients reported in the literature,1,4,21 with three major clinical phenotypes: severe (18%), intermediate (49%), and relatively attenuated (33%). A remarkably large number of patients in the present cohort had an attenuated phenotype, with regression later in life and prolonged survival. The first clinical signs in these attenuated patients were observed at a median age of 10.3 years, and the main symptom was only slightly decreased joint range of motion. Within the first decade of life, these patients did not present typical MPS VI features, their height was only slightly decreased, and diagnosis of MPS VI was frequently made almost 13 years after the appearance of the first symptoms. **Growth is a mirror of health**. James Tanner wrote these important words which since then have become fundamental to the understanding and concept of auxology. For the most rare diseases, we can paraphraph the words that **growing is a reflection of the severity of the disease.** Body height is a very sensitive parameter, reflecting any kind of error in the process of ontogenesis. The average z-score for body height in the present patients was below the WHO reference charts. The lowest deviation from the norm was observed for patients with the attenuated phenotype, while the greatest deviation was noted for patients with the severe

phenotype. The mean z-score for the intermediate phenotype was between these two groups. Individuals with the severe phenotype were not taller than 130 cm, while individuals with an attenuated form of MPS VI grew to a relatively normal height, reaching >145 cm. This shows that body height is directly related to the severity of the disease. Similarly, genotype–phenotype correlation was observed. Missense mutations p.R152W (in homozygosity) and p.Y210C were associated with a relatively attenuated MPS VI phenotype, while heterozygosity for p.R152W mutation yielded an intermediate phenotype. Some missense (p.L72P, p.T92K, and p.Y266S in homozygosity) and nonsense mutations (p.R160X, p.R313X, p.Y513X) were associated with a severe MPS VI phenotype High prevalence of the p.R152W mutation was observed in the whole series (42%). the high prevalence of this mutation observed in the whole series, as well as the Slavic origin of the majority of patients homozygous for this mutation, suggest that p.R152W may be of Slavic, not Lithuanian origin.

Anthropology assessment is manifested not only in the analysis of the natural history of the disease, the creation of phenotypic models and profiles or the creation of patterns of growth. The anthropologist should also use modern technologies in his work. **Publication no 6** (**Bioimpedance analysis as a method to evaluate the proportion of fatty and muscle tissues in progressive myopathy in pompe disease.)** is an example of this. Bioimpedance analysis (BIA) is a noninvasive testing method allowing for accurate analysis of body composition using electrical resistance of various tissues of the body: so-called impedance. It uses the ability of the muscle tissue to conduct electrical current. The body composition assessment method based on electrical bioimpedance analysis is used in medicine, mainly to assess the risk and degree of overweight and obesity, where the fatty tissue to body weight ratio is estimated [38]. The bioimpedance method assesses the tissue components of the body, involving a measurement of electrical resistance in the human body. Resistance varies depending on the composition of the subject's body. The assessment of body composition and the proportion between fatty and muscle tissues is obtained after entering data on age and gender; the device measures body height and weight and performs measurements of resistance and reactance. In study **no 6**, was introduced a completely new, simple to perform, and reliable diagnostic method to assess the progress of the muscle atrophy process in myopathies. Pompe disease is an autosomal-recessive metabolic myopathy, caused by deficiency of the lysosomal acid a-glucosidase. The deficiency of this enzyme leads to an accumulation of glycogen taking place mainly in muscles and leading to their progressive destruction. The spectrum of clinical phenotypes includes infantile-onset form, juvenile form, and late-onset form. In the classic form (infantile onset), the first symptoms appear in the first months of life with hypotonia, muscle weakness, and cardiomegaly. In juvenile forms, the first symptoms, including progressive proximal

and axial muscle weakness, appear between 2 and 5 years of age. The late-onset form has a slower progression. Skeletal muscle involvement is more prominent with a predilection for the lower limbs. Late-onset features include hypotonia and progressive muscle weakness – mainly in respiratory muscles, leading in advanced stages to ventilator-assisted breathing. In Pompe disease, the main affected tissue is muscle tissue – as the disease progresses, this tissue becomes atrophic and is replaced by fatty tissue. This study is innovative because BIA was used for the first time to assess the relative proportions of the fatty and muscle tissues in diseases associated with muscle atrophy, thus enabling the assessment of disease progression and the effectiveness of treatment. The accuracy of BIA is comparable with magnetic resonance [39, 50]. BIA is much cheaper, faster, and easier to perform. The use of BIA to assess the relative proportions of fatty and muscle tissues in diseases associated with muscle atrophy is an innovative method not previously used in myopathies. The results of our study show that there is a correlation between the percentage of body fat by segment and the degree of replacement of the muscle tissue with fatty tissue, as shown in MRI. As the disease progresses, the percentage of body fat increases and reduces the percentage of muscle tissue. Furthermore, it correlates with the age of the patients and the severity of the disease evaluated clinically. The results prove the high sensitivity of the BIA method and suggest great potential for using electric bioimpedance analysis as a cognitive tool enabling the assessment of the proportion of fatty and muscle tissues in progressive myopathies as well as the potential for using the device in diagnostics.

Publication no 7 (**The importance of anthropological methods in the diagnosis of rare diseases)** is a summary of the previous experience of the author in assessing the physical development of patients with rare diseases. The first step in the diagnosis of rare genetic disorders is inspection of the head, face, and body proportions. Knowing all intrapopulation variations of the evaluated feature as well as having a good understanding of the given case (based on a thorough family and social history of the evaluated patient), an anthropologist can assess whether the achieved level of physical development is normal; and if it is not, determine the extent to which it deviates from the norm. Establishing the diagnosis is crucial for initiating treatment and predicting the course of the disease. As a result of underestimating the role of anthropometric methods in describing phenotypes of genetic diseases, phenotypic parameters are typically limited to the patient's height and, possibly, head circumference. Moreover, current diagnostic algorithms tend to focus on molecular and biochemical analyses, while neglecting the relatively simple, non-invasive, and inexpensive anthropometric methods. As a result of such detailed, analytical approach, the patient is less and less often perceived as a whole, with the holistic approach being a fundamental principle of anthropology. Therefore, every major medical research center should have an anthropologic

laboratory with personnel qualified not only to conduct anthropometric measurements but also to accurately interpret the results. Better understanding of the natural history of the disease can be achieved with long-term observations, which help detect and establish patterns of growth and physical development in the evaluated group. Moreover, clinicians typically know only the most characteristic and most common phenotypes for particular genetic diseases, whereas genetic disease phenotypes are likely to have a normal distribution. In which case, it is the anthropologists' role to broaden the scope of what clinicians know and are able to diagnose, by describing also the borderline phenotypes.

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Summary of the most important achievements resulting from the research described in the publication cycle:

- I showed that children with mucopolysaccharidosis were born significantly longer than healthy children in the population. It is an important diagnostic information and a premise for conducting screening tests for mucopolysaccharidosis in children born with birth parameters above 90 percentile.
- For mucopolysaccharidoses type I, II and IVA I have developed a growth pattern that can be used in all MPS I cases; MPS II; MPS IVA for the prognosis of growing and assessment of the severity of the disease.
- I showed that growing is a tool for assessing the effectiveness of treatment.
- I have shown that the key element to describe the rare MPS VI phenotype (mutation p.R152W) was the assesment of growth dynamics within the normal range (genotype-phenotype correlation)
- For the first time I have showed a changing between fat and muscle tissue in metabolic myopathy, facilitating the assessment of the nature of myopathy and its progress in other cases.

5. Other scientific achievements

5.1 Bibliometric analysis (detailed in annex no 6)

<table>
<tr><th colspan="3">Publications</th><th>Number of publications</th><th>IF</th><th>MNISW</th></tr>
<tr><td colspan="3">from Journal Citation Reports</td><td>20</td><td>41,98</td><td>524</td></tr>
<tr><td colspan="2">publications which are scientific achievements submitted for the habilitation procedure:</td><td>6</td><td>8,941</td><td>130</td></tr>
<tr><td colspan="2">publications which are scientific achievements submitted for the habilitation procedure not included in Journal Citation Reports</td><td>1</td><td>0</td><td>5</td></tr>
<tr><td rowspan="3">number of publications from Journal Citation Reports which are not included in scientific achievements submitted for the habilitation</td><td>before obtaining a doctoral degree</td><td>9</td><td>16,22</td><td>224</td></tr>
<tr><td>after obtaining a doctoral degree</td><td>5</td><td>16,82</td><td>165</td></tr>
<tr><td>summary</td><td>14</td><td>33,04</td><td>389</td></tr>
<tr><td colspan="3">publications not included in Journal Citation Reports</td><td>19</td><td>0</td><td>127</td></tr>
</table>

Total number of publications:			**40**	**41,98**	**651**
Hirsch index according to Web of Science			**9**		
Citations/ Citations without self-citations according to Web of Science			**255/232**		
Others informations					
number of scientific projects	**total**				**8**
	national	**grants MNiSW/NCBiR/NFZ**			**4**
		research projects (MPS IVA GALS)			**1**
		IPCZD projects			**2**
	international	**ESPEN grant**			**1**
the number of summaries and conference materials conferences	**total**				**22**
	national				**1**
	international				**21**

5.2. Description of scientific work

5.2.1. Description of scientific work before receiving of doctorate

I gained my first scientific experience during my Master's studies at the Department of Human Biology, Faculty of Biology at the University of Adama Mickiewicza in Poznan, where I wrote my master's thesis on the physical development of children with fragile X syndrome.

In 2007, I have started my job in the Anthropology Laboratory in The Children's Memorial Health Institute in Warsaw. In my daily clinical work I asses:

- Tempo of growing
- Body proportions
- Head and face proportions
- Effectivnes of treatment by controlling the growth of basic development indicators
- Nutrition status (overweight, underweight, other nutritional problems).
- Evaluation of the nutritional status by bioimpedance method.
- Assessment and monitoring of the lower limbs deformity.

In my scientific work I realize research projects carried out at the Anthropology Laboratory. In 2008, I became a member of the research team in the **OLAF research project (PL0080)** implemented from the financial mechanism of the European Economic Area and the Norwegian Financial Mechanism as well as the Ministry of Science and Higher Education. It was the largest research project in Poland aimed at developing blood pressure references for children and adolescents. As part of the project, I carried out research throughout Poland and conducted training for a research team.

Based on data from the project, **standards for body height, body weight and blood pressure for children and adolescents aged 6-18 were made.** In 2010-2012, I was a contractor in the **OLA research project (N R13 0002 06),** financed by the National Center for Research and Development, which was a continuation of the OLAF Project for children aged 3-6 years. The result of the above-mentioned projects were the following publications:

1. Zbigniew Kułaga; Mieczysław Litwin; Maria Małgorzata Zajączkowska; Anna Wasilewska; Aurelia Morawiec-Knysak; **Agnieszka Różdżyńska**; Aneta Grajda; Beata Gurzkowska; Ewelina Napieralska; Katarzyna Barwicka; AnnaŚwiąder; Zespół Badawczy OLAF. Comparison of waist and hip circumferences ranges in children and adolescents in Poland 7-18y of age with cardiovascular risk thresholds – initial tesults of OLAF projects (PL0080). Standardy Medyczne Pediatria 2008 : Vol. 5, Nr 4, s. 473-485.

This study has presented smoothed reference data for waist and hip circumference for Polish children 7-18 y of age for the first time. There was a significant increase in waist circumference in comparison with earlier reports, no different between our results and waist circumference in Canadian children and still lover values in Polish children in comparison with US children population. However, about 30% polish children have waist circumference above cardiovascular risk thresholds.

2. Zbigniew Kułaga; **Agnieszka Różdżyńska**; I. Palczewska; Aneta Grajda; Beata Gurzkowska; Ewelina Napieralska; Mieczysław Litwin; Gr. Badaczy OLAF. Percentile charts for growth and nutritional status assessment in Polish children and adolescents from birth to 18 years of age. Standardy Medyczne Pediatria 2010 : Vol. 7, Nr 4, s. 690-699 Punktacja MNiSW: 6.00

Percentile charts are basic tool for assessment of physical development and early diagnosis of growth and nutritional status disturbances. In this study percentile charts for growth and weight were showed based on actual data from 22 623 health participants.

3. Zbigniew Kułaga; Mieczysław Litwin; M. Tkaczyk; **Agnieszka Różdżyńska**; Katarzyna Barwicka; Aneta Grajda; Anna Świąder; Beata Gurzkowska; Ewelina Napieralska; H. Pan The height-, weight-, and BMI-for-age of Polish school-aged children and adolescents relative to international and local growth references. BMC Public Health 2010 : Vol. 10, 4 March 2010, s. 109 wskaźnik Impact Factor ISI: 2.364, punktacja MNiSW: 27.00

The growth of children is an indicator of health and society's wellbeing. Growth references are useful in monitoring a child's growth, which is a very important part of child care. Poland's growth references are not updated regularly. Although several growth references ranges have been developed in Poland over recent years, sampling was restricted to urban populations of major cities. The aim of this study was to assess how well Polish children match with, or diverge from, regional charts and to compare them with international growth references. In this contemporary sample of Polish school-aged children, distributions of height, weight and BMI differed from those of children from the international growth references. These differences should be considered when using the references. There exist certain limitations to the analysis of height, weight, and BMI z-scores when Polish regional references are used.

I have also begun cooperation with professor Anna Tylki-Szymańska. I have started analysis the physical development of children with various genetic syndromes and metabolic diseases, especially the body and face proportions and characteristic features for rare diseases. The results of this cooperation were several publications. Below are the most important of them:

1. Tylki-Szymańska Anna, Różdżyńska Agnieszka, Jurecka Agnieszka, Marucha Jolanta, Czartoryska B. Anthropometric data of 14 patients with mucopolysaccharidosis I: Retrospective analysis and efficacy of recombinant human alfa-L-iduronidase (laronidase). Mol.Genet.Metab. 2010 Vol. 99 s. 10-17

This study was an initial for other research in the field of analysis of the growth of children with mucopolysaccharidosis, including the doctoral dissertation. Goal of this study was to evaluate growth patterns in terms of body height, weight, head and chest circumference in patients with mucopolysaccharidosis type I (MPS I) without treatment and after enzyme replacement therapy (ERT) with alpha-l-iduronidase (laronidase). Growth patterns were associated significantly with the MPS I at birth. After 96-260 weeks of ERT, patients receiving laronidase compared with group 2 did not show statistically significant improvement. Anthropometric features of patients with MPS I significantly differ from the healthy population. Children with MPS I grew considerably slower, and

differences between healthy and affected children increased with age. In studied patients with MPS I, laronidase did not appear to alter the growth patterns.

2. Agnieszka Jurecka, **Agnieszka Różdżyńska**, Jolanta Marucha, B. Czartoryska, G. Węgrzyn, Anna Tylki-Szymańska. Natural history of Polish patients with mucopolysaccharidosis type VI. Central Eur.J.Med.: 2011 : Vol. 6, Nr 2, s. 163-171

The aim of the study was to describe the natural history, anthropometric features, range of motion (ROM) and molecular characteristics of Polish patients with mucopolysaccharidosis (MPS) VI. Clinical heterogeneity was observed and two major clinical phenotypes of the disease were distinguished, rapidly advancing and relatively attenuated. Two patients developed symptoms early in life presenting with short stature, significant skeletal malformations and other clinical abnormalities. In two other patients, height was only slightly decreased and MPS features developed later in the course of the disease. All patients had similar characteristics at the time of birth but showed significant differences in body proportions when compared with the healthy population. Differences between healthy and affected children increased with age and were reflected in phenotypes. Analysis of ROM showed impairments at multiple joints, although to a various degree in different patients. Restriction in upper extremity ROM was observed since the second year of life, while restriction in lower extremity ROM developed later and influenced stereotype of walking. These limitations intensified with the patients' age, which made self-care more difficult or impossible. The molecular analysis revealed that the milder phenotype may be associated with the R152W mutation, which suggests a specific genotype-phenotype correlation.

3. Jolanta Marucha, Agnieszka Jurecka, **Agnieszka Różdżyńska-Świątkowska**, Anna Tylki-Szymańska. Musculoskeletal manifestations of mucopolysaccharidosis type VI and effects of enzyme replacement therapy. Central Eur.J.Med.: 2012: Vol. 7, Nr 2, s. 154-162

The aim of this study was to describe the musculoskeletal manifestations of mucopolysaccharidosis VI and to assess the effectiveness of enzyme replacement therapy (ERT) with recombinant human arylsulfatase B on the bone and joint involvement in a patient with a severe phenotype of the disease. Before the initiation of ERT, the patient presented with significant range of motion (ROM) limitations at multiple joints. Flexion contractures were noticeable in all joints. After 48 weeks of ERT, improvement in active and passive shoulder flexion, as well as passive elbow and wrist flexion, was noticed. ROM improvements were reflected in patient's enhanced self-care..

4. Agnieszka Jurecka, Z. Krumina, Z. Żuber, **Agnieszka Różdżyńska-Świątkowska**, A. Kłoska, B. Czartoryska, Anna Tylki-Szymańska. Mucopolysaccharidosis type II in females and

response to enzyme replacement therapy. Am.J.Med.Genet. 2012 : Vol. 158A, Iss. 2, s. 450-454

Mucopolysaccharidosis type II (MPS II, Hunter syndrome) is an X-linked lysosomal storage disease caused by a deficiency of iduronate-2-sulfatase (IDS). Two affected girls with moderate and severe forms of MPS II with normal karyotypes and increased urinary dermatan sulphate and heparin sulphate excretion and marked deficiencies of IDS activity are reported. Molecular studies showed that case 1 has a heterozygous mutation c.1568A>G (p.Y523C) associated with almost totally skewed inactivation of the normal maternal X chromosome, and case 2 has a heterozygous deletion that includes exons 1–4 of IDS (minimal deletion range c.1–103_184del). The multi-exon deletion correlated with early onset of the disease and severe phenotype with intellectual disability, whereas the missense mutation was associated with moderate developmental delay. Although genotype–phenotype correlation in MPS II is difficult, gene deletions seem to correlate with more severe clinical manifestation of the disease. Enzyme replacement therapy (ERT) in these two females resulted in disease stabilization in both.

In 2011 I won the competition for the position of a "young scientist" and I was employed as a **scientific assistant** In the Anthropology Laboratory in The Children's Memorial Health Institute in Warsaw. In the years 2012-2013 I was the main contractor in the research project for young scientists entitled: "Impact of enzyme replacement therapy on body height in patients with MPS I and MPS II". The headmaster of this project was professor Anna Tylki-Szymańska. In the results of this project were below publications:

1. **Różdżyńska Agnieszka**, Tylki-Szymańska Anna, Jurecka Agnieszka, Cieślik J.. Tytuł oryginału: Growth pattern and growth prediction of body height in children with mucopolysaccharidosis type II. Acta Paediatr. 2011 Vol. 100 nr 3 s. 456-460

Our goal was to evaluate the level, degree and direction of deviation in the ontogenesis of patients with mucopolysaccharidosis type II (MPS II) in comparison with the healthy population. Material and methods: The anthropometric data of a longitudinal study on 28 patients with MPS II, aged from 0.5 to 21 years, were used to analyse the general growth patterns in terms of height, weight and head circumference. The growth trend was assessed with the straight-line regression model. The mathematical structural growth model was used to evaluate the structure of body height growth. A statistically significant negative growth trend for all features was found. Analysis of development structure revealed an earlier onset of the adolescent growth spurt among healthy boys and a lower current velocity of growth than expected values. During the first 3 years of life, all observed

anthropometric features grew faster than normal. They slowed down by the end of the third year and, in subsequent years, reached lower values when compared with the reference charts. The values obtained from the BTT model showed the structure of body height growth, with particular emphasis on the pubertal spurt, was significantly different from the reference charts

2. Anna Tylki-Szymańska, Agnieszka Jurecka, Z. Zuber, **Agnieszka Różdżyńska**, Jolanta Marucha, B. Czartoryska. Enzyme replacement therapy for mucopolysaccharidosis II from 3 months of age : a 3-year follow-up. Acta Paediatr.: 2012 : Vol. 101 Iss. 1, s. e42-e47

In this study we present a 3-year follow-up of a boy with mucopolysaccharidosis type II (MPS II) who had idursulfase therapy initiated at the age of 3 months and compare his clinical course to his healthy twin brother. Detailed anthropometric features, ultrasound studies of liver and spleen volumes, echocardiography and audiological examinations, psychological tests, joint range of motion (ROM) and skeletal radiographs were monitored. After 3 years of treatment, the patient has not developed any clinical manifestations of MPS II. He did not develop coarse facial features, joint disease, or organomegaly, and his cardiac function remained normal. There were no pronounced signs of dysostosis multiplex on radiographs. The only difference when compared with his healthy twin brother was lower IQ (Termann-Merrill 98 vs. 118) and mild deformity of one vertebrae. This study suggests that early initiation of enzyme replacement therapy may significantly slow or prevent the development of irreversible disease manifestations and therefore modify the natural history of MPS II.

5.2.2. Description of scientific work after receiving of doctorate

In March 2013 I defense my doctoral thesis: Pattern of growth characteristics of physical development in children with mucopolysaccharidosis type I and II (MPS I and MPS II). In July 2013, I became the Head of the Anthropology Laboratory in The Children's Memorial Health Institute in Warsaw.

After getting the doctoral degree I continued to work in OLAF and OLA Projects to analyze the data obtained in these projects. The results were the following publications:

1. **Agnieszka Różdżyńska-Świątkowska**; Zbigniew Kułaga; Aneta Grajda; Beata Gurzkowska; Magdalena Góźdź; Małgorzata Wojtyło; Anna Świąder; Mieczysław Litwin; Gr. Badaczy OLAF i OLA. Height, weight and body mass index references for growth and nutritional status assessment in children and adolescents 3-18 years of age. Standardy Medyczne Pediatria 2013 : Vol. 10, Nr 1, s. 11-21 Punktacja MNiSW: 8.000

The objectives of this study were to update of the percentile charts of height, weight and BMI for children and adolescence in Poland, 3-18 years of age and compare them with local and international growth reference charts. Anthropometric data from 22623 participants health examinations between the ages of 3 and 18 years were used. Percentile charts were constructed using the LMS method with LMS Chart Maker Pro 2.42. We received current and representative for polish population percentile charts which enable basic assessment of physical development and early diagnosis of growth and nutrition status disturbances.

2. Zbigniew Kułaga; **Agnieszka Różdżyńska-Świątkowska**; Aneta Grajda; Beata Gurzkowska; Małgorzata Wojtyło; Magdalena Góźdź; Anna Świąder-Leśniak; Mieczysław Litwin. Percentile charts for growth and nutritional status assessment in Polish children and adolescents from birth to 18 years of age. Standardy Medyczne Pediatria 2015 : Vol. 12, Nr 1, s. 119-135

Objective of this study was presentation of the WHO growth standards for children aged 0-3 years and update of percentile charts of height weight body mass index (BMI) and weight for children and adolescence in Poland 3-18 years of age. LMS parameters according to the WHO growth standards and anthropometric data from 22 623 participants health examinations between the ages 3-18 years were used. Percentile charts were constructed using the LMS method with LMS Chart Maker Pro 2.42. As the result we recesived the growth charts of height, weight, BMI and weight to height based on the WHO growth standards for children aged 0-3 years and current and representative for polish population percentile charts which enable basic assessment of physical development and early diagnosis of growth and nutrition status disturbances. **This study won the Award of Standardy Medyczne for the best original publication in 2015 year.**

In 2014, I became a contractor in the project titled: "Identification of the most common mutations in the GALS gene among Polish patients with MPS IVA and the dependence between phenotype and genotype. **The publication which was as a result of this project is included in the scientific achievement.**

As a result of cooperation with dr hab. Zbigniew Żuber from the Children's Hospital of Saint. Ludwik in Krakow, the following publicatoins were created:

1. Zbigniew Żuber; Agnieszka Jurecka; **Agnieszka Różdżyńska-Świątkowska**; Agata Migas-Majoch; Agnieszka Lembas; Beata Kieć-Wilk; Anna Tylki-Szymańska. Ultrasonographic features of hip joints in mucopolysaccharidoses type I and II. PLoS One 2015 : Vol. 10, Nr 4, s. e0123792

The primary aim of this study was to assess the ultrasonographic features of hip joints in patients with mucopolysaccharidosis (MPS) type I and II in comparison with healthy population. The secondary aims were to correlate these features with clinical measures and to evaluate the utility of ultrasound in the diagnosis of MPS disease. Sixteen MPS I (n = 3) and II (n = 13) patients were enrolled in the present study and underwent clinical and radiological evaluation, and bilateral high-resolution ultrasonography (US) of hip joints. The distance from the femoral neck to joint capsule (synovial joint space, SJS), joint effusion, synovial hyperthrophy, and local pathological vascularization were evaluated. The results were compared to the healthy population and correlated with clinical and radiological measures. Patients with MPS I and II present specific features in hip joint ultrasonography. The data suggests that ultrasonography might be effective in the evaluation of hip joint involvement in patients with MPS and might present a valuable tool in facilitating the diagnosis and follow up of the disease.

2. Zbigniew Żuber; Agnieszka Jurecka; Anna Król-Zdechlikiewicz; Agnieszka Różdżyńska-Świątkowska; Anna Tylki-Szymańska. Bone metabolism in patients with mucopolysaccharidosis type II. Reumatologia 2014 : Vol. 52, Nr 6, s. 354-361

   Which received the Bronze Pen of Rheumatology - award for original publication.

Anthropometric measurements make it possible to calculate and interpret anthropometric and cephalometric proportion indices. Anthropometric proportion indices are used in the diagnostics of numerous diseases, such as the Marfan Syndrome; resistance to thyroid hormone alpha (RTHα); rheumatoid arthritis; or various types of dysplasia. The facial phenotype of Smith-Magenis syndrome includes a wide, prominent jaw (the mandible wider than the maxilla), short, broad-based nose with a full tip. Smith-Lemli-Opitz syndrome is characterized by a narrow forehead, brachycephaly, short palpebral fissures, short nasal bridge, flat face, and retrognathia. Rubinstein-Taybi syndrome is characterized by microcephaly, short and narrow head, narrow palate (maxillary hypoplasia), small mouth, and wide-spaced eyes. Anthropometric measurements and phenotypic profiles may help detect the differences between phenotypes in diseases with the same underlying cause. The Noonan syndrome, Costello syndrome, and cardiofaciocutaneous syndrome all involve Ras/MAPK pathway disturbances, and there are numerous clinical manifestations that these syndromes have in common, with subtle differences between them in terms of anthropometric measurements. In many genetic syndromes it is not only the structure of the face and head that are characteristic, but the shape of the body can also suggest the diagnosis. Altered body proportions are characteristic for Turner syndrome, resistance to thyroid hormone alpha (RTHα), Marfan syndrome, and mucopolysaccharidoses. Like in this study:

- Anna Tylki-Szymańska; Rocio Acuna-Hidalgo; Małgorzata Krajewska-Walasek; Agnieszka Lecka-Ambroziak; Marloes Steehouwer; Christian Gilissen; Han G. Brunner; Agnieszka Jurecka; **Agnieszka Różdżyńska-Świątkowska;** Alexander Hoischen; Krystyna Chrzanowska. Thyroid hormone resistance syndrome due to mutations in the thyroid hormone receptor alpha gene (THRA). Journal of Medical Genetics 2015 : Vol. 52, Nr 5, s. 312-316

Resistance to thyroid hormone is characterised by a lack of response of peripheral tissues to the active form of thyroid hormone (triiodothyronine, T3). In about 85% of cases, a mutation in THRB, the gene coding for thyroid receptor β (TRβ), is the cause of this disorder. Recently, individual reports described the first patients with thyroid hormone receptor α gene (THRA) defects. We used longitudinal clinical assessments over a period of 18 years at one hospital setting combined with biochemical and molecular studies to characterise a novel thyroid hormone resistance syndrome in a cohort of six patients from five families. Findings Using whole exome sequencing and subsequent Sanger sequencing, we identified truncating and missense mutations in the THRA gene in five of six individuals and describe a distinct and consistent phenotype of mild hypothyroidism (growth retardation, relatively high birth length and weight, mild-to-moderate mental retardation, mild skeletal dysplasia and constipation), specific facial features (round, somewhat coarse and flat face) and macrocephaly. We observed a genotype–phenotype correlation, with milder outcomes for missense mutations and more severe phenotypical effects for truncating mutations. Interpretation THRA mutations may be more common than expected. In patients with clinical symptoms of mild hypothyreosis without confirmation in endocrine studies, a molecular study of THRA defects is strongly recommended.

In 2017, I won a competition for the position of **Adjunct** in The Children's Memorial Health Institute in Warsaw.

From 2017 (until 2020) I am the main contractor in two projects financed from the National Health Fund entitled: "Conducting comprehensive epidemiological research on nutrition and nutritional status of Polish society with special consideration of children and adolescents, including identification of risk factor for nutritional disorders , evaluation of the level of physical activity of nutritional knowledge and occurrence of inequities in health within the scope of point 3.1.1 "

I am currently working on the following research topics:

1. Analysis of head and chest circumference index as a determinant of metabolic disease (glutaric acidosis, Alexander's disease and mucopolysaccharidosis).

The effect of this analysis is publication:

- Paulina Pokora, Aleksandra Jezela-Stanek, **Agnieszka Różdżyńska-Świątkowska**, Elżbieta Jurkiewicz Anna Bogdańska, Edyta Szymańska, Dariusz Rokicki, Elżbieta Ciara, Małgorzata Rydzanicz, Piotr Stawiński, Rafał Płoski, Anna Tylki-Szymańska
  Mild phenotype of glutaric aciduria type 1 in polish patients – novel data from a group of 13 cases Metabolic Brain Disease https://doi.org/10.1007/s11011-018-0357-5 Received: 13 June 2018 /Accepted: 25 November 2018

Glutaric aciduria type 1 is a neurometabolic disorder, caused by riboflavin-dependent glutaryl-CoA dehydrogenase deficiency. As its consequence, accumulation of the putatively neurotoxic metabolites (glutaric and 3-hydroxyglutaric acids) in body tissues, but especially within the brain, is observed. Estimated incidence of the disease is 1 in 110,000 newborns, The prevalence however may be higher, depending on a specific ethnic group, and result in phenotypic variation as well. In this paper we present clinical data of 13 patients of Polish nationality. They all present a mild phenotype and clinical course of glutaric aciduria type 1. Based on their clinical data, presented herein, we like to pay attention to the phenotypic and neuroimaging features important for the diagnosis of mild form of this disease. Moreover, we present novel molecular data, which may correlate with such a manifestation.

2. Long - term observation of children born prematurely - when their development begins to be equal to their peers born at the time

3. How to growth children on long term parental nutrition – prediction of pubertal growth spurt of body height.

This is long-term observation and requires the right amount of data. The first analysis were presented on international conference Nutrition&Growth 2018 – poster titled: : *„Prediction of pubertal growth spurt of body height in patients on long-term parenteral nutrition with use of mathematical - structural model*”

4. Bioimpedance analysis in patients with type III Gaucher disease - proportions between fat and muscular tissue in the torso. Hypothesis: dorsal muscle atrophy, the cause of kyphosis in adults.

5. Torso-limb index as a marker for thyroid disorders (including THR alpha, Turner Syndrome).

6. List of publications which are not included in scientific achievements submitted for the habilitation procedure.

6.1. From Journal Citation Reports

1. Paulina Pokora, Aleksandra Jezela-Stanek, **Agnieszka Różdżyńska-Świątkowska**, Elżbieta Jurkiewicz, Anna Bogdańska, Edyta Szymańska, Dariusz Rokicki, Elżbieta Ciara, Małgorzata Rydzanicz, Piotr Stawiński, Rafał Płoski, Anna Tylki-Szymańska
   Mild phenotype of glutaric aciduria type 1 in polish patients – novel data from a group of 13 cases Metabolic Brain Disease https://doi.org/10.1007/s11011-018-0357-5
   Received: 13 June 2018 /Accepted: 25 November 2018
   My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 10%.

2. Anna Tylki-Szymańska; Rocio Acuna-Hidalgo; Małgorzata Krajewska-Walasek; Agnieszka Lecka-Ambroziak; Marloes Steehouwer; Christian Gilissen; Han G. Brunner; Agnieszka Jurecka; **Agnieszka Różdżyńska-Świątkowska**; Alexander Hoischen; Krystyna Chrzanowska. Thyroid hormone resistance syndrome due to mutations in the thyroid hormone receptor alpha gene (THRA). Journal of Medical Genetics 2015 : Vol. 52, Nr 5, s. 312-316
   My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 3%.

3. Paweł Gawliński, Magdalena Pelc, Elżbieta Ciara, S. Jhangiani, Elżbieta Jurkiewicz, Tomasz Gambin, **Agnieszka Różdżyńska-Świątkowska**, M. Dawidziuk, Z.H. Coban-Akdemir, D.L. Guilbride, D. Muzny, J.R. Lupski, Małgorzata Krajewska-Walasek
   Phenotype expansion and development in Kosaki overgrowth syndrome.
   Clinical Genetics 2018 : Vol. 93, Nr 4, s. 919-924
   Paweł Gawliński and Magdalena Pelc contributed equally to this work.
   My contribution is related to: interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 3%.

4. Zbigniew Żuber, Agnieszka Jurecka, **Agnieszka Różdżyńska-Świątkowska**, Agata Migas-Majoch, Agnieszka Lembas, Beata Kieć-Wilk, Anna Tylki-Szymańska
   Ultrasonographic features of hip joints in mucopolysaccharidoses type I and II. PLoS One 2015 : Vol. 10, Nr 4, s. e0123792

My contribution is related to: anthropometric measurements, ,interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 15%.

5. Zbigniew Kułaga, Aneta Grajda, Beata Gurzkowska, Magdalena Góźdź, Małgorzata Wojtyło, Anna Świąder, **Agnieszka Różdżyńska-Świątkowska**, Mieczysław Litwin. Polish 2012 growth references for preschool children. Eur.J.Pediatr. 2013 : Vol. 172, Nr 6, s. 753-761
   My contribution is related to: anthropometric measurements, drafting and revising the article. I declare my contribution to be equal to 5%

6. Zbigniew Kułaga, Mieczysław Litwin, Aneta Grajda, Katarzyna Kułaga, Beata Gurzkowska, Magdalena Góźdź, Hui Pan, the Olaf Study Group, Robert Pietruczuk, **Agnieszka Różdżyńska**, Jan Szpor, Anna Świąder. Oscillometric blood pressure percentiles for Polish normal-weight school-aged children and adolescents. J.Hypertens : 2012 : Vol. 30, Nr 10, s. 1942-1954
   My contribution is related to: anthropometric measurements, drafting and revising the article. I declare my contribution to be equal to 5%

7. Jolanta Marucha, Agnieszka Jurecka, Małgorzata Syczewska, Agnieszka **Różdżyńska-Świątkowska,** Anna Tylki-Szymańska. Restricted joint range of motion in patients with MPS II: correlation with height, age and functional status. Acta Paediatr.2012 : Vol. 101, Iss. 4, s. e183-e188
   My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 10%

8. Jolanta Marucha, Agnieszka Jurecka, **Agnieszka Różdżyńska-Świątkowska**, Anna Tylki-Szymańska. Musculoskeletal manifestations of mucopolysaccharidosis type VI and effects of enzyme replacement therapy. Central Eur.J.Med.: 2012 : Vol. 7, Nr 2, s. 154-162
   My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 15%

9. Agnieszka Jurecka, Z. Krumina, Z. Żuber, **Agnieszka Różdżyńska-Świątkowska**, A. Kłoska, B. Czartoryska, Anna Tylki-Szymańska. Mucopolysaccharidosis type II in females and response to enzyme replacement therapy. Am.J.Med.Genet. 2012 : Vol. 158A, Iss. 2, s. 450-454
   My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 10%

10. Agnieszka Jurecka, Jolanta Marucha, Elżbieta Jurkiewicz, **Agnieszka Różdżyńska-Świątkowska**, Anna Tylki-Szymańska. Enzyme replacement therapy in an attenuated case of mucopolysaccharidosis type I (Scheie syndrome): a 6.5-year detailed follow-up. Pediatr.Neurol.: 2012 : Vol. 47, Nr 6, s. 461-465
    My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 10%

11. Anna Tylki-Szymańska, Agnieszka Jurecka, Z. Zuber, **Agnieszka Różdżyńska**, Jolanta Marucha, B. Czartoryska. Enzyme replacement therapy for mucopolysaccharidosis II from 3 months of age : a 3-year follow-up. Acta Paediatr.: 2012 : Vol. 101 Iss. 1, s. e42-e47
    My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 15%

12. Agnieszka Jurecka, **Agnieszka Różdżyńska**, Jolanta Marucha, B. Czartoryska, G. Węgrzyn, Anna Tylki-Szymańska. Natural history of Polish patients with mucopolysaccharidosis type VI. Central Eur.J.Med.: 2011 : Vol. 6, Nr 2, s. 163-171
    My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 25%
13. **Różdżyńska Agnieszka**, Tylki-Szymańska Anna, Jurecka Agnieszka, Cieślik J..
    Growth pattern and growth prediction of body height in children with mucopolysaccharidosis type II. Acta Paediatr. 2011 Vol. 100 nr 3 s. 456-460
    My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 60%

14. Kułaga Zbigniew, Litwin Mieczysław, Tkaczyk M., **Różdżyńska Agnieszka**, Barwicka K., Grajda Aneta, Świąder A., Gurzkowska Beata, Napieralska Ewelina, Pan H. The height-, weight-, and BMI-for-age of Polish school-aged children and adolescents relative to international and local growth references. BMC Public Health 2010 Vol. 10:109 s. 1-26
    My contribution is related to: anthropometric measurements, interpretation of data, drafting and revising the article. I declare my contribution to be equal to 15%

15. Tylki-Szymańska Anna, **Różdżyńska Agnieszka**, Jurecka Agnieszka, Marucha Jolanta, Czartoryska B. Anthropometric data of 14 patients with mucopolysaccharidosis I:

Retrospective analysis and efficacy of recombinant human alfa-L-iduronidase (laronidase). Mol.Genet.Metab. 2010 Vol. 99 s. 10-17

My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 30%

6.2. Not included in Journal Citation Reports

1. Zbigniew Kułaga; Mieczysław Litwin; Aneta Grajda; Beata Gurzkowska; Anna Świąder-Leśniak; **Agnieszka Różdżyńska-Świątkowska**; Magdalena Góźdź; Małgorzata Wojtyło. Normy rozwojowe wysokości i masy ciała, wskaźnika masy ciała, obwodu talii i ciśnienia tętniczego dzieci i młodzieży w wieku 0-18 lat. Standardy Medyczne Pediatria 2015 : Vol. 12, Supl. 1, s. 3-44

   My contribution is related to: anthropometric measurements, interpretation of data, drafting and revising the article. I declare my contribution to be equal to 10%

2. Zbigniew Kułaga; **Agnieszka Różdżyńska-Świątkowska**; Aneta Grajda; Beata Gurzkowska; Małgorzata Wojtyło; Magdalena Góźdź; Anna Świąder-Leśniak; Mieczysław Litwin. Siatki centylowe dla oceny wzrastania i stanu odżywienia polskich dzieci i młodzieży od urodzenia do 18 roku życia. Standardy Medyczne Pediatria 2015 : Vol. 12, Nr 1, s. 119-135

   My contribution is related to: anthropometric measurements, interpretation of data, drafting and revising the article. I declare my contribution to be equal to 25%

3. Anna Świąder-Leśniak; Zbigniew Kułaga; Aneta Grajda; Beata Gurzkowska; Magdalena Góźdź; Małgorzata Wojtyło; **Agnieszka Różdżyńska-Świątkowska**; Mieczysław Litwin. Wartości referencyjne obwodu talii i bioder polskich dzieci i młodzieży w wieku 3-18 lat. Standardy Medyczne Pediatria 2015 : Vol. 12, Nr 1, s. 137-150

   My contribution is related to: anthropometric measurements, drafting and revising the article. I declare my contribution to be equal to 5%

4. Zbigniew Żuber; Agnieszka Jurecka; Anna Król-Zdechlikiewicz; **Agnieszka Różdżyńska-Świątkowska**; Anna Tylki-Szymańska. Metabolizm kostny u pacjentów z mukopolisacharydozą typu II. Bone metabolism in patients with mucopolysaccharidosis type II. Reumatologia 2014 : Vol. 52, Nr 6, s. 354-361

   My contribution is related to: anthropometric measurements, interpretation of data, drafting and revising the article. I declare my contribution to be equal to 10%

5. **Agnieszka Różdżyńska-Świątkowska**, Zbigniew Kułaga, Aneta Grajda, Beata Gurzkowska, Magdalena Góźdź, Małgorzata Wojtyło, Anna Świąder, Mieczysław Litwin, Grupa Badaczy OLAF i OLA, Robert Pietruczuk, Jan Szpor. Wartości referencyjne wysokości, masy ciała i wskaźnika masy ciała dla oceny wzrastania i stanu odżywienia dzieci i młodzieży w wieku 3-18 lat. Standardy Med. Pediatria. 2013 : Vol. 10, Nr 1, s. 11-21
   My contribution is related to: anthropometric measurements, interpretation of data, drafting and revising the article. I declare my contribution to be equal to 55%

6. Zbigniew Kułaga, Aneta Grajda, Beata Gurzkowska, Magdalena Góźdź, Małgorzata Wojtyło, Anna Świąder, **Agnieszka Różdżyńska-Świątkowska**, Mieczysław Litwin, Grupa Badaczy OLA. Siatki centylowe do oceny ciśnienia tętniczego dzieci i młodzieży w wieku 3-18 lat. Standardy Med. Pediatria. 2013 : Vol. 10, Nr 1, s. 22-30 *My contribution is related to: anthropometric measurements, interpretation of data, drafting and revising the article. I declare my contribution to be equal to*
   mój udział procentowy szacuje na 5%

7. Agnieszka Jurecka, V. Opoka-Winiarska, J. Szczepański, **Agnieszka Różdżyńska**, Jolanta Marucha, Anna Tylki-Szymańska; Kostno-stawowa manifestacja mukopolisacharydozy typu VI (choroby Maroteaux-Lamy'ego).Reumatologia 2011 : Vol. 49, Nr 4, s. 288-293
   My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 10%

8. Kułaga Zbigniew, Litwin Mieczysław, Wójcik P., Jakubowska-Winecka Anna, Grajda Aneta, Gurzkowska Beata, Napieralska Ewelina, **Różdżyńska Agnieszka**. Przyczyny i nasilenie zgonów dzieci wiejskich w latach 1999-2006 - porównanie z dziećmi miejskimi. Medycyna Ogólna 2010 T. 16 nr 1 s. 63-75
   My contribution is related to: anthropometric measurements, interpretation of data, drafting and revising the article. I declare my contribution to be equal to 5%

9. Anna Świąder, **Agnieszka Różdżyńska**, Katarzyna Popińska, Elżbieta Arasimowicz, Janusz Książyk. Ocena rozwoju fizycznego dzieci z zespołem krótkiego jelita na podstawie wybranych parametrów antropometrycznych. Pediatr.Endocrinol. Diab.Metab. 2010 : Vol. 16, Nr 4, s. 284-288

My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to: 25%

10. Agnieszka Jurecka, **Agnieszka Różdżyńska**, Jolanta Marucha, B. Czartoryska, Anna Tylki-Szymańska. Mukopolisacharydoza typu VI (choroba Maroteaux-Lamy'ego) - opis przypadku. Pediatr.Med.Rodz. 2010 : Vol. 6, Nr 2, s. 151-155

My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 25%

11. Jurecka Agnieszka, **Różdżyńska Agnieszka**, Marucha Jolanta, Czartoryska B., Tylki-Szymańska Anna. Choroba Maroteaux-Lamy'ego (mukopolisacharydoza typu VI): obraz kliniczny, diagnostyka i leczenie. Pediatr.Pol. 2010 T. 85 nr 4 s. 305-310

My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 25%

12. Jurecka Agnieszka, **Różdżyńska Agnieszka**, Marucha Jolanta, Czartoryska B., Tylki-Szymańska Anna. Polscy pacjenci z chorobą Maroteaux-Lamy'ego (mukopolisacharydozą typu VI).Pediatr.Pol. 2010 T. 85 nr 4 s. 311-319

My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 25%

13. Jurecka Agnieszka, Marucha Jolanta, **Różdżyńska Agnieszka**, Czartoryska B., Tylki-Szymańska Anna. Przypadek mukopolisacharydozy typu VI (choroby Maroteaux-Lamy'ego) - charakterystyka kliniczna. Pediatr.Pol. 2010 T. 85 nr 4 s. 399-406

My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 15%

14. Kułaga Zbigniew, **Różdżyńska Agnieszka**, Palczewska I., Grajda Aneta, Gurzkowska Beata, Napieralska Ewelina, Litwin Mieczysław, Grupa Badaczy OLAF , Siatki centylowe wysokości, masy ciała i wskaźnika masy ciała dzieci i młodzieży w Polsce - wyniki badania OLAF. Standardy Med. 2010 T. 7 nr 4 s. 690-699

My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 25%

15. Arasimowicz Elżbieta, Pietrucha Barbara, **Różdżyńska Agnieszka**, Heropolitańska-Pliszka Edyta. Różnice w budowie głowy i ciała dzieci z zespołem ataksja-telangiektazja. Pediatr.Endocrinol.Diab.Metab. 2009 T. 15 nr 3 s. 196-202

My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 15%

16. Kułaga Zbigniew, Litwin Mieczysław, Zajączkowska M., Wasilewska A., Tkaczyk M., Gurzkowska Beata, Świąder A., **Różdżyńska A**., Napieralska Ewelina, Grajda Aneta, Barwicka K., Zespół Badawczy OLAF Regionalne różnice parametrów antropometrycznych oraz ciśnienia tętniczego uczniów w wieku 7-18 lat. Probl.Hig.Epidemiol. 2009 T. 90 nr 1 s. 32-41

My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 5%

17. Kułaga Zbigniew, Litwin Mieczysław, Wójcik P., Jakubowska-Winecka Anna, Grajda Aneta, Gurzkowska Beata, Napieralska Ewelina, Barwicka K., **Różdżyńska A.**, Wiśniewski T. Aktualne trendy zewnętrznych przyczyn zgonów dzieci i młodzieży w Polsce. Probl.Hig.Epidemiol. 2009 T. 90 nr 3 s. 332-341

wkład pracy kandydata: krytyczne zrecenzowanie artykułu pod kątem istotnej zawartości intelektualnej

mój udział procentowy szacuje na: 5%

18. **Różdżyńska Agnieszka**, Cieślik J., Latos-Bieleńska A., Nowak M..

Fragile X syndrome and physical development course. W: Somatic development, physical fitness and health status of rural children and adolescents, pod red. H. Popławskiej. Wyd. Josef Pilsudski University of Physical Education in Warsaw Faculty of Physical Education in Biała Podlaska 2009 s. 263-276

My contribution is related to: anthropometric measurements, interpretation of data, statistic analysis, drafting and revising the article. I declare my contribution to be equal to 60%

19. Zbigniew Kułaga, Mieczysław Litwin, M. Zajączkowska, A. Wasilewska, A. Morawiec-Knysak, **A. Różdżyńska**, Aneta Grajda, Beata Gurzkowska, Ewelina Napieralska, Katarzyna Barwicka, Anna Świąder, Zespół Badawczy OLAF. Porównanie wartości obwodów talii i bioder dzieci i młodzieży polskiej w wieku 7-18 lat z wartościami referencyjnymi dla oceny ryzyka sercowo-

naczyniowego - wyniki wstępne projektu badawczego OLAF (PL0080).Standardy Med. Pediatria, 2008 : Vol. 5, Nr 4, s. 473-485

My contribution is related to: anthropometric measurements, interpretation of data, drafting and revising the article. I declare my contribution to be equal to 5%

7. Conference abstracts and congress reports publish in abstracts books with IF:

1. Anna Tylki-Szymańska; Agnieszka Różdżyńska-Świątkowska; Jolanta Marucha; Agnieszka Ługowska Enzyme replacement therapy for mucopolysaccharidosis type II from 3 months of age: 9-year follow up.
   Journal of Inherited Metabolic Disease 2016 : Vol. 39, Supl. 1, s. S275 Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM).
   Rome, Italy, 2016.08.06-2016.08.09

2. Agnieszka Różdżyńska-Świątkowska; Elżbieta Jurkiewicz; Anna Tylki-Szymańska Pompe disease - the proportion of fatty andmuscle tissues as an indicator of progression and severity of the disease.
   Journal of Inherited Metabolic Disease 2016 : Vol. 39, Supl. 1, s. S65
   Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM) :
   Rome, Italy, 2016.08.06-2016.08.09

3. Agnieszka Różdżyńska-Świątkowska; Joachim Cieślik; Anna Tylki-Szymańska
   Using mathematical - structural model in prediction of pubertal spurt in patients with MPS I and MPS II.
   Journal of Inherited Metabolic Disease 2016 : Vol. 39, Supl. 1, s. S191
   Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM)
   Rome, Italy, 2016.08.06-2016.08.09

4. Agnieszka Różdżyńska-Świątkowska; Anna Tylki-Szymańska. Anthropologist's contribution supporting diagnosis of rare diseases.
   Journal of Inherited Metabolic Disease 2015 : Vol. 38, Supl. 1, s. S67
   Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM).
   Lyone, France, 2015.08.01-2015.08.04

5. Anna Tylki-Szymańska; Agnieszka Różdżyńska-Świątkowska; Jolanta Marucha; A. Kułpanowicz; A. Tulebayeva; Agnieszka Jurecka.
   Molecular analysis of 22 patients with mucopolysaccharidosis IVA from Poland, Belarus and Kazakhstan identifies 6 novel GALNS mutations.
   Journal of Inherited Metabolic Disease 2015 : Vol. 38, Supl. 1, s. S242
   Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM): Lyone, France, 2015.08.01-2015.08.04

6. Agnieszka Różdżyńska-Świątkowska; Anna Tylki-Szymańska MPS II - patient's profile - objective evaluation of the body stature in patient who started idursulfase treatment presymptomatically at the age of 3 months.
   Journal of Inherited Metabolic Disease: 2015 : Vol. 38, Supl. 1, s. S242
   Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM): Lyone, France, 2015.08.01-2015.08.04

7. Agnieszka Różdżyńska-Świątkowska; Agnieszka Jurecka; Jolanta Marucha; Anna Tylki-Szymańska
   Birth body length and weight at birth in patients with MPS I, MPS II and MPS VI.
   Journal of Inherited Metabolic Disease 2014 : Vol. 37, Supl. 1, s. S129
   Annual Symposium of the Society for the Study of Inborn Errors of Metabolism (SSIEM)Innsbruck, Austria, 2014.09.02-2014.09.05

8. Katarzyna Olszewska; Elżbieta Banaś; Małgorzata Janusz; Maciej Jaworski; Agnieszka Różdżyńska-Świątkowska; Joanna Friedman-Gruszczyńska; Mikołaj Danko; Katarzyna Popińska; Janusz Książyk.
   Bone mineral density in children with intensinal failure weaned off parenteral nutrition. Journal of Pediatric Gastroenterology and Nutrition 2014 : Vol. 58, Supl. 1, s. 516 47th Annual Meeting of the European Society for Paediatric Gastroenterology, Hepatology and Nutrition. Jerusalem, Israel, 2015.06.09-2014.06.12

9. Agnieszka Różdżyńska-Świątkowska, Anna Tylki-Szymańska. MPS IVA - patient's profile - objective evaluation of the body stature. J.Inherit.Metab.Dis. 2014 : Vol. 37, Supl. 1, (SSIEM) Annual Symposium of the Society for the Study of Inborn Errors of Metabolism; 2-5 September 2014; Innsbruck, Austria

10. Agnieszka Różdżyńska-Świątkowska, A. Jurecka, Jolanta Marucha, Anna Tylki-Szymańska Birth body length and weight at birth in patients withMPS I, MPS II and MPS VI. J.Inherit.Metab.Dis. 2014 : Vol. 37, Supl. 1, s. S129
    SSIEM 2014: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism; 2-5 September 2014; Innsbruck, Austria

11. Janusz Książyk, Aleksandra Żyła, Agnieszka Różdżyńska-Świątkowska. Growth of children on home parenteral nutrition. Abstract 169; 2nd International Conference On Nutrition And Growth: Barcelona, Spain, 30 Januray - 1 February, 2014

12. Katarzyna Olszewska, Elżbieta Banaś, Małgorzata Janusz, Maciej Jaworski, Agnieszka Różdżyńska-Świątkowska, Joanna Friedman-Gruszczyńska, Mikołaj Danko, Katarzyna Popińska, Janusz Książyk; Bone mineral density in children with intensinal failure weaned off parenteral nutrition.
    J.Pediatr.Gastroenterol. Nutr.2014 : Vol. 58, Supl. 1, s. 516; 47th Annual Meeting of the European Society for Paediatric Gastroenterology, Hepatology and Nutrition; 9-12 June, 2014; Jerusalem, Israel

13. Agnieszka Różdżyńska-Świątkowska, Z. Żuber, A. Jurecka, Anna Tylki-Szymańska. Comparison of growth patterns of patients with Mucopolysaccharidosis type II who started enzyme replecement therapy before 6 years of age and patients who were naive to ERT.
    J.Inherit.Metab.Dis. 2013 : Vol. 36, Supl. 2, s. S301-S302 ICIEM 2013 12th International Congress of Inborn Errors of Metabolism, 3-6 September, 2013, Barcelona

14. Jolanta Marucha, Agnieszka Jurecka, Małgorzata Syczewska, Agnieszka Różdżyńska-Świątkowska, Anna Tylki-Szymańska. Restricted joint range of motion in MPS II patients: correlation with age and height. J.Inherit.Metab.Dis. 2012 : Vol. 35, Suppl. 1, s. S90

15. A. Jurecka, L. Cimbalistiene, N. Gusina, V. Malinova, Agnieszka Różdżyńska-Świątkowska, A. Gołda, A. Kulpanovich, G. Kaldenovna Abdilova. Natural history, incidence and prevalence rates of mucopolysaccharidosis type VI in Central and Eastern Europe.
    J.Inherit.Metab.Dis. 2012 : Vol. 35, Nr 1 Supl., s. S90 Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Birmingham, UK, 4-7 September 2012

16. A. Jurecka, Z. Krumina, Z. Żuber, Agnieszka Różdżyńska-Świątkowska, A. Kłoska, B. Czartoryska, Anna Tylki-Szymańska. Mucopolysaccharidosis type II in femals and response to enzyme replacement therapy
    J.Inherit.Metab.Dis. 2012 : Vol. 35, Nr 1 Supl., s. S146 Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Birmingham, UK, 4-7 September 2012

17. A. Jurecka, E. Piotrowska, L. Cimbaistiene, N. Gusina, Agnieszka Różdżyńska, B. Czartoryska, K. Ounap, G. Węgrzyn, Anna Tylki-Szymańska. Genotype-phenotype correlation in mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)
    J.Inherit.Metab.Dis. 2011 : Vol. 34, Supl. 3, s. S213 Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Geneva, Switzerland, 30 August - 2 September 2011

18. Anna Tylki-Szymańska, A. Jurecka, Agnieszka Różdżyńska, Jolanta Marucha, B. Czartoryska. Enzyme replacement therapy in a 3-month-old boy with presymptomatic MPS II: 3-year follow-up.
    J.Inherit.Metab.Dis. 2011 : Vol. 34, Supl. 3, s. S209 Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Geneva, Switzerland, 30 August-2 September 2011

19. A. Jurecka, Jolanta Marucha, Agnieszka Różdżyńska, B. Czartoryska, Anna Tylki-Szymańska. Efficacy of recombinant human arylsulfatase B (galsulfase) on restrictedrance of motion and activittes of daily living in patients with Maroteaux-Lamy syndrome (mucopolysaccharidosis type VI): improvement after 24 weeks of treatment.
    J.Inherit.Metab.Dis. 2010 : Vol. 33, Supl. 1, s. S152. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Istanbul, Turkey, 31 August - 3 September 2010

20. A. Jurecka, Agnieszka Różdżyńska, Jolanta Marucha, B. Czartoryska, Anna Tylki-Szymańska. Natural history, detailed anthropometric data and joint range of motion of patients with Maroteaux-Lamy syndrome (mucopolysaccharidosis type VI). J.Inherit.Metab.Dis.2010 : Vol. 33, Supl. 1, s. S136 Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Istanbul, Turkey, 31 August - 3 September 2010

21. Jurecka Agnieszka, Różdżyńska Agnieszka, Marucha Jolanta, Czartoryska B., Tylki-Szymańska Anna. Growth patterns of 14 patients with mucopolysaccharidosis I: retrospective analysis and efficacy of recombinant human alfa-L-Iduronidase (Laronidase). Mol.Genet.Metab. 2009 Vol. 98 nr 1-2 s.62

8. Scientific projects

2017- 2020 Contractor in two projects financed from the National Health Fund entitled "Conducting comprehensive epidemiological studies on the diet and nutritional status of Polish society with special consideration of children and adolescents, including the identification of risk factor for nutritional disorders, assessment of physical activity level of nutritional knowledge and health inequalities in in point 3.1.1 "

Contracts: No. 6 / 113.1.1aINPV2O17/ 100/630 and 6/1/3.1.1a/NPZ/2017/100/631

2014 Contractor in the project "Identification of the most common mutations in the GALS gene among Polish patients with MPS IV A and establishing the dependence of the clinical picture on the genotype" Grant from Association for patients with Mucopolysaccharidosis (MPS) and Rare Diseases; KRS number 0000060517 (http://chorobyrzadkie.pl/)

2012-2013 Main contractor in the The Children's Memorial Health Institute research projekt for young scientists "Evaluation of the impact of enzyme replacement therapy of patients with MPS I and MPS II on body height".

"M3/12 ZAD.BAD. The development of young scientists"

2010-2012 Contractor - member of the research team in the OLA project (NR 13 0002 06), financed from the National Center for Research and Development. "Development of blood pressure norms, body height and body mass and BMI in children aged 3-6 in Poland

2009-2010 Contractor - member of the research team in the OLAF project (PL0080), financed from the Financial Mechanism of the European Economic Area and the Norwegian Financial Mechanism and the Ministry of Science and Higher

Education "Development of blood pressure norms for children and adolescents in Poland".

09.2010 Participation in the project "Impact of nutritional status on the results of hospitalization in children" within the ESPEN Grand Project - contractor

9. Received awards and distinctions

2015 Award of Standardy Medyczne for the best original publication in 2015 year.

Zbigniew Kułaga; Agnieszka Różdżyńska-Świątkowska; Aneta Grajda; Beata Gurzkowska; Małgorzata Wojtyło; Magdalena Góźdź; Anna Świąder-Leśniak; Mieczysław Litwin. Percentile charts for growth and nutritional status assessment in Polish children and adolescents from birth to 18 years of age. Standardy Medyczne Pediatria 2015 : Vol. 12, Nr 1, s. 119-135

2014 Bronze Pen of Rheumatology Award for original publication:

Zbigniew Żuber; Agnieszka Jurecka; Anna Król-Zdechlikiewicz; Agnieszka Różdżyńska-Świątkowska; Anna Tylki-Szymańska. Bone metabolism in patients with mucopolysaccharidosis type II. Reumatologia 2014 : Vol. 52, Nr 6, s. 354-361

1-2.04.2011 10th International Symposium on Lysosomal Storage Diseases – poster *"Assessment of physical growth in patients with mucopolysaccharidosis type II"* third place in the competition for the best poster in the category of clinical research (Madrid, Spain)

10. Oral presentation on national and international scientific conferences

22-23.11.2018 Agnieszka Różdzyńska-Świątkowska. *Dynamic of growth and anthropological features in MPS II.* Central and East Europe MPS II Expert meeting, Zagreb, Croatia

13-15.09.2017 Agnieszka Różdżyńska-Świątkowska, Elżbieta Jurkiewicz, Anna Tylki-Szymańska. *Bioimpedance analysis as a method to evaluate the proportion of fatty and muscle tissuesin progresive myopathy.* XLVI National Scientific Conference of the Polish Anthropological Society Szczecin, Poland

2.07 -6.07. 2015 Agnieszka Różdżyńska-Świątkowska. *The role of an anthropologist in the diagnosis of rare diseases.* XIII International Conference of Rare Diseases "Rare Diseases Together We Cross Borders - oral presentation: Białobrzegi; Poland

8.04-11.04.2015 Agnieszka Różdżyńska-Świątkowska. *Anthropology as an useful tool supporting diagnosis of rare disease.* International Congress of Anthropological Science. Ankara, Turky

17-19.09.2013 Agnieszka Różdżyńska-Świątkowska. Physical development of patients with a rare metabolic disease - mucopolysaccharidosis. XLIV National Scientific Conference of the Polish Anthropological Society. Warsaw Poland.

11-12.09.2009 Agnieszka Różdżyńska, Zbigniew Kułaga, Mieczysław Litwin. Comparison of the waist and hip circumferences of Polish children and youth aged 7-18 with reference values for cardiovascular risk assessment - preliminary results of the OLAF research project. XLII National Scientific Conference of the Polish Anthropological Society. Łódź, Poland

10-12.06.2008 Agnieszka Różdżyńska. *Fragile X syndrome and the course of physical development.* 5th International Scientific Conference: Determinants of the development of children and adolescents in rural areas, Biała Podlaska, Poland.

11. Didactic and popularizing achievements

11.1. Posters on national and international scientific conferences

7.03-09.03.2019 Agnieszka Różdżyńska-Świątkowska, Joanna Żydak, Mikołaj Danko, Honorata Kołodziejczyk, Anna Świąder-Leśniak, Janusz Książyk. Poster: Familial marasmus syndrome of unknown cause.6th International Conference on Nutrition & Growth, Valencia, Spain

1-3.03.2018 Agnieszka Różdżyńska-Świątkowska, Magdalena Durda –Masny, Aleksandra Żyła-Pawlak, Janusz Książyk. Poster: Prediction of pubertal growth spurt of body height in patients on long-term parenteral nutrition with use of mathematical - structural model. 5th International Conference on Nutrition & Growth, Paris, France

2.03-4.03.2017 Agnieszka Różdżyńska-Świątkowska, Anna Tylki-Szymańska. Poster1: "The usefulness of anthropological methods in the assessment of the physical development in patients with inborn errors of metabolism".
Aleksandra Żyła-Pawlak, Agnieszka Różdżyńska – Świątkowska, Katarzyna Popińska, Janusz Książyk. Poster 2: "Long-term parental nutrition influences on growth in children"
4th International Conference on Nutrition and Growth, Amsterdam, Holand;

6.09-9.09.2016 Agnieszka Różdżyńska-Świątkowska, Joachim Cieślik, Anna Tylki-Szymańska. Poster 1: "Using mathematical – structural model in prediction of pubertal spurt in patients with MPS I and MPS II"
Agnieszka Różdżyńska-Świątkowska, Elżbieta Jurkiewicz, Anna Tylki-Szymańska. Poster 2: "Pompe Disease – the proportion of fatty and muscle tissues as an information of progression and severity of the disease" Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Rome, Italy

21.09 – 25.09.2015 Agnieszka Różdżyńska-Świątkowska, Elżbieta Jurkiewicz, Anna Tylki-Szymańska. Poster "Bioimpedance analysis as a method to evaluate the proportion of fatty and muscle tissues in progressive myopathy in Pompe disease. 44th European Muscle Conference, Warsaw, Poland

09.09-11.09.2015 Agnieszka Różdżyńska-Świątkowska, Anna Tylki-Szymańska. MPS II – patient's Profile – Obiective evaluation of the body stature in patient who started idursulfate treatment presymptomatically at the age of 3 months. Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Lyon, France.

01.09-05.09.2014 Agnieszka Różdżyńska-Świątkowska, Anna Tylki-Szymańska Poster "MPS IV A – patient's profile - objective evaluation of the body stature"

Agnieszka Różdżyńska-Świątkowska, Agnieszka Jurecka, Anna Tylki Szymańska: Poster "Birth body length and weight at birth in patients with MPS I, MPS II and MPS VI"

Annual Symposium of the Society for the Study of Inborn Errors of Metabolism. Innsbruck, Austria

30.01-2.02.2014 Aleksandra Żyła, Agnieszka Różdżyńska-Świątkowska, Katarzyna Popińska, Anna Świąder , Janusz Książyk. Poster: Growth of children on home parental nutrition. 2nd International Conference of on Nutrition and Growth. Barcelona, Spain.

3 – 6.09. 2013 Agnieszka Różdżyńska-Świątkowska, Zbigniew Żuber, Agnieszka Jurecka, Anna Tylki-Szymańska. Poster: "Comparison of growth patterns of patients with mucopolysaccharidosis type II who started enzyme replacement therapy before 6 years of age and patients who were naïve to ERT".12th International Congress of Inborn Errors of Metabolism. Barcelona, Spain.

3-6. 09. 2011 Agnieszka Różdżyńska-Świątkowska, Agnieszka Jurecka, Jolanta Marucha, Anna Tylki-Szymańska. Poster: „Compatison of growth pattern of patients with MPS type I and II. 18th European Study Group on Lysosomal Diseases - Workshop Helsinki, Finland.

1-2.04.2011 Agnieszka Różdżyńska, Barbara Czartoryska, Joachim Cieślik. Poster "Assessment of physical growth in patients with mucopolysaccharidosis type II" 10th International Symposium on Lysosomal Storage Diseases – **third place in the competition for the best poster in the category of clinical research.** Madryt, Spain.

31.05.2008 Agnieszka Różdżyńska, Andrzej Wiśniewski, Elżbieta Arasimowicz. Poster pt: *Proportion of the lower limbs in the Turner syndrome - preliminary report.* VII Conference - Progress in the assessment of disorders of physical development: shortage of growth - in chronic diseases and arthritis. Warsaw, Poland

11.2. Didactic

- In 2008, training of research groups in the OLAF project "Development of norms of arterial pressure of children and adolescents in Poland" entitled: Methodology of anthropometric measurements.

- Since 2010 (regularly during student internships) - Conducting lectures and exercises for students of the Medical Academy as part of student internships at the Department of Paediatrics, Nutrition and Metabolic Diseases: "Anthropometric assessment of nutritional status"
- 11/2014 Agnieszka Różdżyńska-Świątkowska. Rare metabolic diseases: - biochemical background, clinical picture and physical development of patients, lectures for students and members of the Polish Anthropological Society as part of the cycle "New research areas in physical anthropology - from genotype to phenotype" Poznań, Poland

- 21/11/2015 Agnieszka Różdżyńska-Świątkowska, Elżbieta Jurkiewicz, Anna Tylki-Szymańska. Lecture: Bioimpedance analysis as a method to evaluate the problem of fatty tissue in Pompe disease. XV Workshop Genzyme Sanofi: Therapeutic perspectives and 20 years of experience in the treatment of lysosomal storage diseases. Warsaw Poland

- In 2018 training for research groups in projects "Conducting comprehensive epidemiological studies on the diet and nutritional status of Polish society with special consideration of children and adolescents, including the identification of risk factor for nutritional disorders, assessment of physical activity level of nutritional knowledge and health inequalities in in point 3.1.1 "

12. Membership in international and national organizations and scientific societies

Since 2016, a member of the Polish Anthropological Society.

13. Reviewing publications in magazines

• BMC Medical Genetics 2018 year 1 publication

• Journal of Pediatric Rehabilitation Medicine 2015 year 1 publication

• Molecular Genetics and Metabolism 2014 year 1 publication

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