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A Thirteen-Year-Old Girl with Cornelia de Lange Syndrome – The First Case Described in Litigation

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Abstract

AIM: The aim of this paper is to accentuate importance of the expert witness role, occupational medicine specialist, in litigation. The OM specialist proved his importance in broad spectrum of administrative court proceedings, assessment of working capacity, and different type of claims.

CASE REPORT: Here is presented a case of a 13-year-old girl suffering Cornelia de Langue syndrome. The occupational medicine specialist concludes that the girl's claim toward the Ministry of Demography, Family and Social Affairs is legitimate. Expert witness representing the above ministry came to conclusion that the patient is not heavily disabled due to her mobility and regular school attendance. Due to hearing aid her hearing is satisfactory and she is capable of dressing and feeding herself. In 2012, the girl was diagnosed with Langer-Giedion syndrome 8q23.3-q24.13 deletions and due to the development of molecular diagnosis only in 2015 RAD 21 gene deletion was discovered and she was correctly diagnosed. On examination, occupational medicine specialist found the patient suffering heavy deformations of locomotor system, small hands and feet, genua valga, and flat feet. The hearing is severely impaired in right and moderately in her left ear. Her mother states that she stopped soiling bed at the age of 8 and at present when going to toilette during night she becomes disoriented, sleeps badly, and screams. In the morning, she is not capable of preparing her own food. The patient needs to be examined by endocrinologist, her body is covered in exostoses and she heavily depends on other people.

CONCLUSION: The patient is heavily disabled and in need of help for essential functioning at least until completion of secondary education.

Introduction

Cornelia de Langue syndrome (CdLS) is a severe genetic disorder characterized by multi systemic malformations [1]. CdLS is due to pathogen variants in NIPBL, SMC1A, SMC3, RAD 21, and HDAC8 genes, which belong to the cohesion pathway. As a consequence, the patients suffer from behavioral problems, urinary incontinence, feeding difficulties, sleep disturbance, growth difficulties, facial dimorphisms, tone and motor problems, and other abnormalities [2]. Sensorineural hearing loss and high arched palate are frequent [3]. In affected patients with severe hearing loss, cochlear implantation is recommended [4].

The aim of this paper is to accentuate the importance of expert witness's assessment, because patient's future depends on expert's expertise. In this case, according to the occupational medicine specialist, for a 13-year-old girl the help and assistance of her mother is essential, not only because of her future progress but also to prevent relapsing into her previous, more severe state. The girl frequents the seventh grade of regular elementary school and is aided in only two classes, gymnastics, and musical education. With hearing aid, her hearing is satisfactory, speech is comprehensible and she uses orthopedic shoes. Because of her partial independence, at first sight, this patient does not appear to be severely disabled. After a very superficial assessment by the Ministry of Demography, Family and Social Affairs experts, misinterpreting very strict rules that do not apply to the patient, they came to the conclusion that heavy disability does not exist, thus making the patient illegible for the state disability allowance that she was receiving from 2011 to 2016.

The mother, as a legal representative of the underage girl took the above ministry to court. The occupational medicine specialist is chosen as an expert witness in the legal proceeding.

Case Report

This case presents a 13-year-old girl with atypical CdLS. The diagnosis is under the International classification list of diagnoses Q 93.7. There is a microdeletion of the eight chromosomes (8q23.3-q24.13) that influences protein synthesis and cohesion, which is important for gene expression and DNA reparation.

At first, in 2012, the patient had been diagnosed a Langer-Giedion syndrome [5]. After the advancements made in molecular and genetic diagnostics in the Republic of Croatia, in 2015, she was diagnosed with CdLS [6].

The mother, the legal representative of the underage girl, took the Ministry of Demography, Family and Social Affairs to court after her daughter's disability allowance was suspended in 2016.

In this legal proceeding, an occupational medicine specialist was used as an expert witness.

To gather all the relevant data, in the presence of her mother, the OM specialist examined the girl personally.

The patient appeared timid, pale in the face, and obese. Her height is 149 cm, and she weighs 69 kg, body mass index = 31. Blood pressure was 105/70 mm Hg. She wears spectacles and hearing apparatus in both ears. She achieves satisfactory visual acuity, for the right eye 0.8 and for the left eye 1.0. with correction. With hearing aid in both ears, her hearing of normal speech is satisfactory.

She is relatively communicative and answers simple questions while always looking at her mother.

She communicated that she frequented the seventh grade of elementary school, that she walks to school by herself 15 min each way and at present is not an A grade student but achieves C and B grades. The patient is able to dress and undress on her own.

However, her independence is rather limited, the girl is very shy, and the mother informs us that she stopped soiling her bed at the age of 8, she becomes disoriented in the night coming back to bed from toilette and sometimes walks into a wall and hurts herself. She sleeps badly and often has nightmares, so she screams. In the morning, she cannot prepare her own breakfast, nor can she manage a gas cooker.

She is pale due to heavy periods lasting up to 30 days, so the additional endocrine examination is needed. She had her first period at the age of ten and developed some body hair already at 2 years old.

It is evident that the patient has small arms and exostosis of fingers, small feet with the second and the third toe fused together (Figure 1), and she wears specially adapted orthopedic shoes.

Further, the patient has Grade IV flat feet and genua valga. The hip rotation is limited to half a perimeter.

The audiometry test demonstrated severe sensorineural damage for the right and moderate for the left ear.

Discussion

From the above description, it is easy to conclude that there exist severe locomotor apparatus deformations and a severe loss of hearing in the right, moderately in the left ear. The patient has not stopped growing and developing yet and which causes new problems almost every day. Due to very long and heavy periods, there is a danger of severe anemia; thus, additional examination is urgent as there is endangering of a severe anemia.

Figure 1: Exostosis manus, flat feet with short fingers, the second and the third toe fused together, and genua valga

The main problem in the assessment of this patient is the interpretation of rules that determine disability, as well as approaching this patient as an individual.

In their assessment, the ministry's council of expert witnesses used the Book of Rules [7] in which Article 38. Section 2 states that one's health is severely damaged if one is not capable of independent walking with the orthopedic tools, not fed through gastrostoma or other feeding tubes, not capable of maintaining personal hygiene and physiological needs, not capable of independently dressing, not capable of receiving and memorizing information, incapable of establishing social contacts with other persons, which makes one completely dependent on the care of others.

According to the above mentioned book of rules, superficially looking, the patient has not got a heavy disability. However, "incapacity to maintain personal hygiene and physiological needs" is not adequately interpreted by the ministry's experts.

If the patient must get up in the night to use the toilette (she soiled the bed until the age of eight) and is not capable to find her way back – that means that without the essential help of other person (her mother), the capacity of basic physiological needs and personal hygiene becomes at least questionable. In fact, relapsing, i.e., soiling the bed is the most likely result.

According to the Article 44 of the same Book of Rules, the essential need for help of the other person exists, when the person cannot independently satisfy basic life demands, because the person cannot independently walk in or out of doors, take food, dress and undress, keep personal hygiene, and perform basic physiologic needs.

This patient cannot independently feed herself, because she is incapable of cooking or heating up her own food, as an injury is a very likely possibility. Therefore, the council of ministry's experts had to take all of the above into consideration as it is obvious that the patient is helpless and suffers heavy invalidity.

The same confirms the Book of Rules and regulations of the Methodology of Expertise [8]. It states that chromosomal aberrations make a 100% of body damage; therefore, a heavy invalidity does exist. If adding up separately, ones again 100% of body damage includes: Conditions that lead to partial dependency on other person (60%), total loss of hearing 70–90% feet deformities 30%, limited hip rotation 30%, and all added it comes up to 100%.

The patient also demonstrates anxiety in social interaction with unfamiliar persons [9], which is another symptom of the above-mentioned syndrome. Further psychological examination for this time was not planned.

Occupational medicine expert witness concluded that on the basis of both Books of Rules (the first used by the ministry's council of experts), and the second which he used personally, the heavy invalidity, without any doubt does exist. The patient suffers changes on molecular level, changes in "micro" world that influences her whole body in "macro" world. Rules and regulations are written in general, and sometimes they cannot describe or comprehend all atypical changes that originate, for instance, on the genetic level, which, on the other hand, experts must seriously take into consideration.

Conclusion

It should be clear that this is a case of a heavy disability and that the patient has to be regularly reassessed, before enrolling in secondary education and on completing it.

Once again, as already many times in the past, Occupational Medicine proved its importance in litigation processes [10].

Occupational medicine specialist came to conclusion already now that the girl will be incapable of a large number of occupations in the future. Therefore, in this legal process, it confirms her disability at present.

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