

CENTRE FOR CHILDREN AND ADOLESCENTS, UNIVERSITY CLINICAL CENTER, PEDIATRIC CLINIC LJUBLJANA, SLOVENIA

Majda Oštir



SLOVENIA



Population

2021: 2.108.977; counting 2002: 1.964.036 Population density: 104/km² Main city: Ljubljana – 260.000 people Official language: Slovenian, Italian, Hungarian Independence from Yugoslavia was declared d 25.6.1991 and recognized in 1992 EU entrance 1 May 2004, € GDP 2018 estimate total: usd 75.967 bilion – per capita :USD 36

SLOVENIA











3 CF CENTERS 1 PEDIATRIC 2 ADULTS



UKC Ljubljana, Pediatric clinic, Department for lung diseases

78

https://www.ecfs.eu/ecfspr



27

UKC Ljubljana, Clinical department for pulmonology and lung diseases



8

Clinic for lung diseases and allergy Golnik

COVID 19 IN OUR CF CENTRE

Prevention activities, CF newsletters, pamphlets about prevention, telephone calls

All outpatient visits (every 3 months) and annual checkups (1 per year) were done.

Data from December 2021

12 (16.6 %) patients recovered from covid 19, all of them unvaccinated before infection

Thirty-five (48,6 %) were fully vaccinated (n=72). Their mean age was 14 ± 6.4 years.

COVID-19 vaccination in children and adolescents with cystic fibrosis - a single centre experience. A.Zver¹, M. Praprotnik¹, M. Aldeco¹, D. Lepej¹, S. Šetina Šmid¹, J. Rodman Berlot¹, U. Krivec¹. *Department for Pulmonology, University Children's Hospital Ljubljana, University Medical Centre Ljubljana, Ljubljana, Slovenia*

PROPORTION OF CHILDREN (<18 YEARS) AND ADULTS (\geq 18 YEARS), BY COUNTRY. PATIENTS ALIVE ON 31/12/2019. Poland 863 (70.11) (1

Children (<18 years) number (%) 57(50.44)

Adults (≥18 years) number (%)

| | · · · | |
|--------------------|---------|---------|
| Poland | 863 | 368 |
| | (70.11) | (29.89) |
| Portugal | 191 | 173 |
| | (52.47) | (47.53) |
| Romania | 229 | 13 |
| | (94.63) | (5.37) |
| Russian Federation | 2526 | 855 |
| | (74.71) | (25.29) |
| Serbia | 134 | 67 |
| | (66.67) | (33.33) |
| Slovak Republic | 134 | 187 |
| | (41.74) | (58.26) |
| Slovenia | 57 | 56 |
| | (50.44) | (49.56) |
| Spain | 1112 | 1292 |
| | (46.26) | (53.74) |
| Sweden | 264 | 438 |
| | (37.61) | (62.39) |
| Switzerland | 427 | 568 |
| | (42.91) | (57.09) |
| Turkey | 1752 | 240 |
| | (87.95) | (12.05) |
| Ukraine | 169 | 41 |
| | (80.48) | (19.52) |
| United Kingdom | 4515 | 6026 |
| | (42.83) | (57.17) |
| Total | 24041 | 26447 |
| | 21012 | |

REVIEW AND ASSESSMENT OF DISEASE INDICATORS IN CHILDREN AND ADOLESCENTS WITH CYSTIC FIBROSIS IN SLOVENIA

Original research article, Slovenian Medical Journal; In press

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DEMOGRAPHIC CHARACTERISTICS

78 patients, of whom 56% (44/78) were male patients.

The mean age of the patients was 13.2 years (SD 6.4 years), ranging from 1.2 years to 25.5 years.

The proportion of patients under 18 years of age was 70.5% (55/78).

Two patients after lung transplantation in 2019 and 2020



GENETICS AND DIAGNOSIS

Mutation of both alleles of the CFTR gene was identified in all patients.

19 different mutations were detected.

The most common F508del, followed by G542X (10.3%), R1162X (9%) and CFTRdel2.3 (3.8%).

As many as 88.5% of patients are carriers of at least one F508del mutation, and in 64.1% of patients, the mutation is present on both alleles.

The median age at diagnosis was 7 months in our patients (IQR 1–24 months)

NUTRITION



The median z-BMI value was -0.2 kg/m2

In 64.0% (32/50) under 18 years of age, the z-value of BMI was less than 0.0 kg/m2 (from 0.1 to 0.4 kg/m2)

The mean BMI in patients older than 18 years was 21.4 kg/m2

LUNG FUNCTION



At least one spirometry measurement was performed by 63 patients older than 6 years.

The average FEV1% was 91.3% and the average FVC% was 94.1% (from 102% to 79%)



MICROBIOLOGY

19.2% (15/78) of all patients had chronic P. aeruginosa infection.

Chronic infection was detected in 7.7% (4/52) of patients under 18 years of age.

2 patients with Mycobacteria abscessus

- 4 patients with MRSA
- 2 patients with Achromobacter species



TREATMENT WITH CFTR MODULATORS IN 2020

Lumakaftor / ivacaftor

Five children (F508 homozygotes) began receiving dual CFTR modulator drug, lumacaftor/ivacaftor (mean age 7.8 years, FEV1% before drug initiation 75.7%).

Eleksakaftor / tezakaftor / ivakaftor

In August 2020, in the program of orphan medicine we started introducing a triple CFTR modulating drug and initially introduced the drug to nine patients with advanced diseases who were carriers of the F508del mutation.

Following the official registration of the drug in November 2020, we continued to introduce the drug to other patients for whom the drug is indicated. By the end of 2020, we introduced the treatment to sixteen patients, which represents 20.5% of all our patients (mean FEV1% before the introduction of the drug 80.7%, LCI before drug administration 15.1).

Folowing protocol 0, 1m, 3m, 6m, 9m, 12m, 24 m

Comparison of indicators of diseases of children and adolescents with cystic fibrosis in Slovenia in 2020 with the European average for 2019.

For comparison with other European centers, we used data from the annual report of the European register of cystic fibrosis patients for 2019.

| Disease indicator | Slovenija (2020) | EU register |
|--|------------------|---------------------|
| | | (2019) |
| Age at diagnosis (month), mediana (IQR) | 7 (1-24) | 4 (1-32) |
| FEV1% (6-18 let), average (SD) | 98,0 % (16,4 %) | 90,4 % |
| BMI z-value (younger from od 18 y), median (IQR) | -0,2 (-0,7-0,2) | -0,2 (-1,0- 0,5) |
| Chronic infections Pseudomona aeruginosa (younger from18 y), % | 7,7 % | 12,8 % |

WORKING WITH AND FOR PATIENTS

- regular monthly CF team meeting
- availability on telephone 8 hours per day (also on email)
- CF newsletter
- booklets for individuals with CF
- Yearly educational day for health professionals working with individuals with CF
- Yearly educational day

for individuals with CF and their families



INFORMATION FOR PATIENTS



re dinke a sistôrio fibrizio je, še posebej v dosobeni tevnejše kot v ostalih družinat. Otrementve imajo Clations fibroza in dullevno zdravje ply na dullevno zómeje in dobrobit posametri undecija za ostično fibruzu skupaj cli ti priedil ano dve zidensi, in alter

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vita ekro zase preprečuje negativna lahko štiti pred nastarikom manejših

nist section of the sector cem pri apoprijemenju

totkov CF centre Pediatrične stroke UNC Ljubijene 2018

9 and v CF centra Pediatrible Minika LINC devili Të bolcikov (KTE devilo). V povoredju ao bil etert 12.3 ete. Bhov mediani FEV1 (pijulina funccija) je znatal (H.7% in indexa erre mese (HM), 43 standardinega oddona, Indeset odstotov inkov je bio krostično kolongiranih z betarlji. Pseudomoraz as Devel adaption in is melo a CF poverano alackor n. En bornk je mel prezijene pjuže. Povprečni vrednu e funkcije in incekae belenie nase sta tik ob povprečni struh bornkos primečjive starost v Evropalem regist na odationi forozo Firski stat. 40.10.





V VSAKDANJEM ŽIVLJENJU

Mikrobi so povsod, lahko pa storite marsikaj, da zmanjšate tveganje za obolenje. Nasveti v nadaljevanju so namenjeni ozaveščanju, da boste zase znali sprejemati najboljše odločitve. Ta priporočila temeljijo na Smernicah za preprečevanje in nadzor okužb pri cistični fibrozi (CF), ki jih je izdala fondacija Cystic Fibrosis Foundation. Primarni cilj smernic je pomagati ljudem s CF, da ohranijo in zaščitijo svoje zdravje.

Ohranjajte varno razdaljo 2 metrov

Mikrobi se lahko razpršijo do 2 metrov daleč s kapljicami, ki se sproščajo v zrak, kadar kašljate ali kihate. Skušajte ohranjati razdaljo vsaj 2 metrov od drugih s CF ali kogar koli drugega, ki ima prehlad, gripo ali okužbo v vseh okoljih tako izven in še zlasti znotraj prostorov, kot sta šola, delovno mesto ali zdravstvena ustanova. Ljudje s CF, ki ne živijo skupaj, naj se izogibajo aktivnostim, pri katerih bi prišlo do tesnega stika z drugimi osebami s CF ali s komer koli, ki je bolan. Izogibati se je treba naslednjim aktivnostim:

CYSTIC FIBROSIS FOUNDATION

• rokovanju, objemanju ali poljubljanju

- vožnji v istem avtomobilu
- bivanju v isti hotelski sobi
- obiskovanju istih vadb v fitnesu.

Umivajte si roke



Mikrobe lahko dobite in jih širite naprej, ko se dotaknete nečesa, kjer so mikrobi že prisotni, kot sta kljuka ali ograja, in se potem dotaknete svojih oči, nosu ali ust. Umijte si roke z milom in vodo ali jih očistite z gelom za roke na osnovi alkohola. Tudi člane svoje družine in prijatelje spodbujajte, naj imajo čiste roke.



CONCLUSION

•Waiting for official registration for Eleksakaftor / tezakaftor / ivacaftor for children 6-11 years old (already preparing patients)

•Approved national program for neonatal screening in 2022

•Aprroved program for improvements in pediatric CF center in 2022

•New edition of Handbook for children and adolescents with cystic fibrosis 2022 – in press

•Challenges in the transition process



