Childhood Experiences and Reproductive Decision-Making in the Era of CFTR-Modulator Therapy

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Parenthood and Reproductive Decision-Making

German CF Registry, 2023 (1)

Brief overview	2000	2005	2010	2015	2020	2022	2023
Age in years; Median[1]	14	16	18	20	21	22	23
Proportion of adults (≥ 18 years) in %	36.3	45.9	51.3	56.7	58.7	60.2	61.0
Maternities in the reporting year	8	9	16	23	37	57	63
Paternities in the reporting year	4	3	7	8	18	18	16
Age at death in years; median	21	26	28	31,5	35	37	38,5
Transplant patients in the reporting year[1]	9	36	33	34	33	9	5

(1) Nährlich, L. et al, (2024). Deutsches Mukoviszidose-Register: Berichtsband 2023.

(2) Jain, R., et al. (2022). Pregnancy in cystic fibrosis: Review of the literature and expert recommendations. Journal of cystic fibrosis, 21(3), 387–395.

Parenthood in People with CF

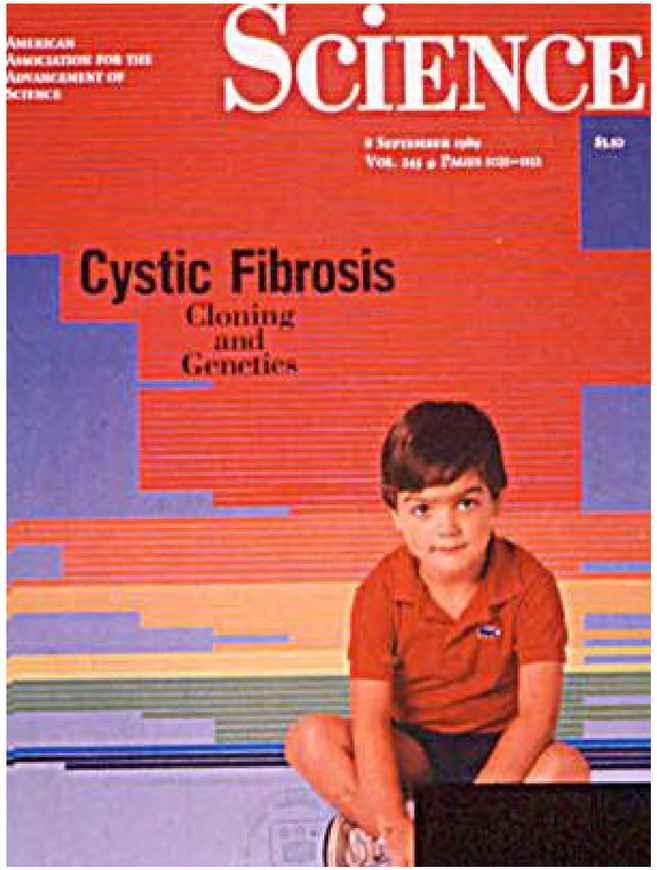
- Increased rates of pregnancy and fatherhood among pwCF over the past 30 years, accelerating around 2020 (1)
- Made possible by **better overall health** + **quality of life** due to improved therapy + **CFTR modulators that increase female fertility** + better **financial status** due to better education and higher employment rates (1,2)

A realistic chance to have and provide for children

1. **Demographic transition** Life goals of adulthood partnership & family
2. **Better health** + Higher fertility + better education Reaching these goals is realistic
3. **successful pregnancies** w/o significant loss in lung function Increase in pregnancies GER 63, US 619 in 2023

Jahresbericht des Deutschen Mukoviszidose-Registers 2021, Jain & Taylor-Cousar 2021, Jacob et al. 2021, Kazmerski et al., 2022, McMullen et al., 2006; Sawyer, 2007; Nash et al., 2020; Ah

Diagnosis and Genetic Testing across stages of family planning and pregnancy



Before pregnancy: Carrier-Scrrening of the partner in affected families, (+/- Pre-implantation)

During pregnancy: “Prenatal Diagnosis” NIPT week 8 since 2019 in GER, CVS week 16, Amniocentesis week 20 (since ~1989s)

After birth: Newborn Screening since 2017 in GER Metabolic +/- genetic testing within the first days of life. Symptomatic children with meconium ileus, failure to thrive, recurrent infections, usually before the age of 1 year

time

Cover of Science 1989; 245(4922), celebrating the „Identification of the cystic fibrosis gene” by Riordan, Rommens, Kerem, et al.

Identititfication & Reproductive Decisions in Rare Genetic Conditions

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- Attitudes toward selective reproduction are dependent on the way in which participants view their condition as part of their identity (Boardman & Hale 2018)
- Individuals with CF and other rare genetic diseases often appreciate their lives, but hesitate to have a child with the same condition (König & Reinsch 2024, Reinsch & König SS&M under review).

Our research is situated at the intersection of two discussions

1. The so-called **'expressivist objection'** (Boardman 2014) that testing is a moral insult to people living with the condition tested and a de-valuation of their life
2. People's experience with illness or **'disability expertise'** (Hartblay 2020) – and how this shapes identification. In the case of CF an identity that is liminal – neither healthy nor ill – and shaped by constant biographical revisions (Reinsch Medical Anthropology in press)

➢ What are the reproductive imaginaries of pwCF, how do they anticipate the future, and with what consequences? (Anderson 2010; Povinelli 2011; Ginburg & Rapp 2020)

	Interview	Gend-er	Age	Modulator Therapy	Social Support	Paren-tal Status	Child-ren n	Part-ner Test-ing	Prenatal Diag-nostic	Would have a Child w/ CF
CF6	Ellen	F	35	Yes	-	Parent	2	+	NIPT	Yes
CF13	Martin	M	31	Yes	-			+	+	Yes
CF9	Martina	F	26	Yes	-	Parent	1	-	-	Yes
CF10	Tim	M	24	Transplant	-			-	-	Yes
CF14	Anke	F	57	Yes	-			-	-	Yes
CF5	Clara	F	32	Transplant	-/+			+		No
CF1	Peter	M	56	Yes	+	Parent	3	+	-	No
CF2	Magda-lena	F	32	Yes	+			+	-	No
CF3	Anna	F	30	Yes	+	Parent	1	+	-	No
CF4	Franziska	F	31	Yes	+			+	Uncertain	No
CF8	Konrad	M	38	Yes	+	Parent	1	+	+	No
CF11	Ralf	M	33	Yes	+			+	+	No
CF12	Maria	F	31	Yes	+	Parent	2	+	-	No
CF15	Jan	M	23	No	+			+	+	No
CF7	Alice	F	38	Yes	+	Parent	1	-	NIPT	Yes
n=15		6M, 9W	34y	13/15	6/9	7 (47%)	1,5	9/15	6/15	6/15

Reproductive Decision-Making

Would pwCF test for CF and if so, when - before, during or after pregnancy?

→ Most would test their partner

→ Once pregnant, women would carry the pregnancy to term

? testing as anticipatory practice aiming to prevent, prepare or preempt?

Do pwCF want to avoid having a child with CF?

→ This seems to depend on the support they received from their social environment during their upbringing

→ We find an inverse correlation between supportive parenting and the willingness to have a child with CF

The Moral Resoning Behind the Decision

15 People with CF

6 Experienced lack of support

9 Experienced support

Existential insult:
I was constantly, and still am, controlled and insulted, being told that I can't do anything and can't get anything done. (woman, 26, one child)

Prove to oneself:
With his parents, when I casually mentioned that I wished for children, they said, 'Well, first take care of yourself.' (women, 35, two children)

Chance to do better:
They say that you should do better than your parents, that would be the chance, whether my child has CF or not. (men, 23)

Abandonment: My biological father couldn't deal with (my CF) and he apparently told my mother he didn't want a child with something wrong with it. (woman, 35, two children)

Scapegoat: The result of all my therapy is simply that they always used my illness for everything they couldn't do themselves. (woman, 57)

Family conflict: After the divorce of his parents (that he attributed partly to his CF) this triangle relationship between my dad, my brother and me really didn't do any good. It was very rough. (men, 31)

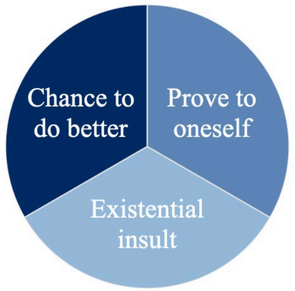
Nurturing: Because I saw the effort my mother put in, which I also want to put in, so that my child would be well of. (woman, 32, one child)

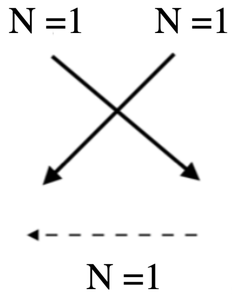
Autonomy: My parents supported me well, both financially and with everything else that was needed. (woman, 38, one child)

Experienced care as standard:
Because I, as a CF patient, don't want to have a CF child. It is too much. I wouldn't be able to handle that. (woman, 34)

Carefreeness:
A life without CF is simply more carefree. And that carefree nature, that's what it's all about. (woman, 32)

Partnership priorities:
So if we had said we're not having a child with CF, it would have been more of a decision for us, and not against the child. (woman, 32, one child)





Child with CF

Child without CF

Boardman, F. K., & Hale, R. (2018). How do genetically disabled adults view selective reproduction? Impairment, identity, and genetic screening. Molecular genetics & genomic medicine, 6(6), 941–956.

König, A., & Reinsch, S. (2024). “I am happy to be alive, but I prefer to have children without my chronic disease”: chronically ill persons’ views on reproduction and genetic testing for their own condition. New Genetics and Society, 43(1).

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Comments & Questions

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