Panel ABSTRACT

Panel 17: Care and transition in chronic and rare conditions

Organizer: Malgorzata Rajtar, Eva-Maria Knoll

Abstract: Developments in medicine and medical technologies have significantly reduced mortality in many chronic and rare conditions. People with these conditions - many of which start in childhood - are now living longer, even into adulthood. Novel gene therapies enable living with a chronic disease and give people hope for a cure. In this dynamic healthcare landscape, practices of giving and receiving care and self-care are in transition; they are "in situ enactments of long-term transformations of human relationships, informed by fantasies of futurity and post hoc perceptions of experience" (Cook and Trundle 2020:178).

In this panel, we will interrogate the relationship between care and transition(s) in the globally changing healthcare landscape characterized by competing political, medical, socio-economic, migrational, and religious agendas and concerns. Focusing on chronic and rare conditions, we seek to critically examine how practices of care and self-care are "unsettled" (Cook and Trundle 2020) and/or "unsettling" (Murphy 2015) in the context of transition; how transition can be redefined to go beyond biomedical care; and how anthropologists may illuminate temporal, spacial, and sensorial dimensions of caring and transition(s). We are also interested in contributions which attend to non-human actors in the transition process.

We invite ethnographically grounded contributions from medical anthropologists as well as scholars in science and technology studies, critical disability studies, feminist ethics, and critical global health, among others. We are also open to engaged submissions that draw from scholarship in graphic medicine and/or sensorial studies.

SESSION SCHEDULE

18.09.2025 | Slot 1| 4-0-6

Valentine Gourinat, Francesca Incagli, Alice Colombo, Martina Lanza, Ruta Butkeviciene, Lina Danuseviciene, Yaël Slaghmuylder, Emelien Lauwerier, Lisa Gittel, Aggée Célestin Lomo Myazhiom, David Le Breton, Hélène Dollfus: Transitions to Adulthood for Youth with Rare Genetic Eye Diseases: A Sociological Analysis of Care,

Support, and Quality of Life

Nicoletta Diasio: Parents in between: medical transition and intergenerational relationships in France

Valentina Turén, Marcela González-Agüero, Constanza Quezada, Ivonne Vargas: Living against time: Care, chronicity and legal struggles of patients with Spinal Muscular Atrophy in Chile

Clara Fabian Therond: The changing nature of hope: following its journey across pathways of personalised medicine care in late stage cancer treatment settings

Yiting (Eliza) Ge: Imagining the Future Elsewhere: Hope, Transnational Care, and Rare Disease Patients in China

Meng Kou: Endless pain, hope that is not understood, eternal mental strength -Ethnography of migrant family with young adult patient of sickle cell disease in France

Zuzanna Kuffel: "Complicated" Veins in "Complicated" Women: Pelvic Venous Insufficiency as Condition Multiple

Ilaria Lesmo: Generatively Unsettling Care: On Distrust and Failure in Transition to Imagine New Forms of Care

Claire Mavis: Diagnosed and Dumped: hEDS Care and the Diagnostic Moment

Sabina Vassileva: The Loop in Transition: Attending to Hormonal Intra-actions in Automated Diabetes Care

Syeda Shajeela Shawkat: The Social Body in Focus: Constructing Personhood through Intimacy

Malgorzata Rajtar: In limbo. Entanglements of care, ethics, and law in transition processes of people with rare diseases

SESSION PAPERS

Transitions to Adulthood for Youth with Rare Genetic Eye Diseases: A Sociological Analysis of Care, Support, and Quality of Life

Valentine Gourinat, Francesca Incagli, Alice Colombo, Martina Lanza, Ruta Butkeviciene, Lina Danuseviciene, Yaël Slaghmuylder, Emelien Lauwerier, Lisa Gittel, Aggée Célestin Lomo Myazhiom, David Le Breton, Hélène Dollfus

The transition from childhood or adolescence to adulthood is often complex and marked by various constraints, fears, and challenges (Le Breton, 2007). This is especially true for children with disabilities or rare chronic diseases, as becoming an independent adult often requires additional preparation and tailored support (Lewi-Dumont, 2015). These

conditions cause significant stress and anxiety for families (Faivre & al., 2025), hindering long-term life projection. In response to this, some families overprotect their child, which can unintentionally slow the development of skills for independence (Gagnon & al, 2020) and hamper the possibility of living an adult life far from the family.
In addition, young people with rare diseases or disabilities face unique difficulties during this transition between childhood and adulthood (Bathelt, 2019). They may struggle to relate to conventional models of adulthood, particularly concerning employment opportunities and independent living in an ableist society that is not designed for their needs (Savin & al., 2024). What strategies and behaviours do these young people or their families develop to anticipate and deal with this specific issue?
This presentation draws on a sociological study focused on the quality of life of young people with a rare genetic eye disease causing visual impairment. In-depth interviews with young people, their families, caregivers, and support workers will explore the challenges, resources, and limitations involved in transitioning to adulthood. The analysis will highlight key concerns such as employment prospects and independent living, identifying needs and strategies developed throughout their life course.

Parents in between: medical transition and intergenerational relationships in France

Nicoletta Diasio

The paper focuses on the transition from paediatric to adult medical services for young people aged 15-22 living with type 1 diabetes in France. Based on two research projects (Growing up with a chronic illness, for an anthropology of uncertainty, IUF, and Age and medical transitions in the care trajectories of 12-22 year olds, IRESP-INSERM) and three sets of documents (ethnographic observations in health centers; 51 interviews and observations with young patients and their families; 45 interviews with health professionals), we analyze the transformation of the role of parents and how their participation in the management of the disease and intergenerational relations are renegotiated. The transition between services redistributes the fonctions and the positions of all social actors: while it primarily affects young patients, it also contributes to giving parents a new role, placing them in a system of double binds. On the one hand, health professionals ask them to remain involved in their children's care, and on the other, they are urged to 'cut the cord' and let their children become independent. This double injunction is not new in French society, where the institution, be it educational or medical, has the role of interposing itself between the generations in order to facilitate children's access to citizenship. How do health professionals and parents cope with these paradoxical requirements and the changes brought about by the medical transition? And how do young patients deal with this?

Living against time: Care, chronicity and legal struggles of patients with Spinal Muscular Atrophy in Chile

Valentina Turén, Marcela González-Agüero, Constanza Quezada, Ivonne Vargas

Spinal Muscular Atrophy (SMA) is a rare neurodegenerative disease that progressively

limits the ability to walk, speak, swallow, and breathe. In Chile, there are approximately 300 known cases mainly affecting children and young people. Currently, the healthcare system does not guarantee appropriate access to diagnosis and pharmacological treatment, forcing patients and their families to resort to legal action to claim their right to health and obtain medications that could prolong their lives with less disability.
This paper argues that time, far from being a neutral or linear dimension, is a key category for understanding how people affected by SMA confront, negotiate, and resist multiple forms of exclusion within the Chilean healthcare system. Through 12 months of fieldwork with adolescents and young people with SMA, we have observed how time emerges as a contested, polysemic category in constant tension among various actors who shape their life trajectories. Temporality intersects with the urgency of diagnosis and treatment and the slowness of institutional responses; with biomedical projections and bureaucratic delays; with daily rhythms marked by illness and the rapid progression of physical deterioration.
Time also transforms care dynamics, implicating caregivers and families who must adapt to the changes imposed by the disease. Thus, the temporality of this group is not limited to the biomedical progression of the illness or the rapid pace of pharmaceutical innovation but is continuously redefined through interaction with their broader social environment—marked by structural precarity and local resistance strategies aimed at sustaining these lives.

The changing nature of hope: following its journey across pathways of personalised medicine care in late stage cancer treatment settings

Clara Fabian Therond

Developments in personalised medicine practices are transforming treatment and care in the late-stage cancer setting, in the UK National Health Service (NHS). As care moves away from a one size fits all approach to treating people according to their molecular profile there is hope to transition cancer from a life-threatening disease into a chronic condition. This paper examines different genetic testing technologies coupled with promissory personalised therapeutics, such as immunotherapy to explore how they not only challenge understanding of the body, and relationships between human and non-human actors, but cultivate a novel discourse, practice, and method of hope across and beyond care pathways.
Discussion is based on comparative ethnographic fieldwork conducted in the ovarian and colorectal cancer NHS treatment setting between 2020-2021, in a large tertiary hospital at the forefront of delivering personalised medicine care. Discussion focuses on the relationship between care and hope, as it travels across research and clinical pathways, within two different personalised therapeutics, and between those delivering and receiving personalised medicine, to understand how it shapes subjective experiences in new and (un)familiar ways.
By following hope this paper exposes its materialities: how it shapes expectations, competes with suffering, dovetails disappointment, and transforms time into a technology of care. Indeed, by attending to hopes competing temporal, moral, cultural, political, and clinical dynamics this paper not only seeks to understand its (less)visible affect on people providing and receiving personalised medicine but concludes by asking what hope is in

this changing landscape of care.

Imagining the Future Elsewhere: Hope, Transnational Care, and Rare Disease Patients in China

Yiting (Eliza) Ge

This paper examines how the future is imagined and lived by patients with life-threatening rare diseases in contemporary China. Focusing on individuals who currently lack treatment options in China but are waiting for clinical trial results in the U.S., the study explores how these patients perceive their personal futures in relation to China's vision of national progress, its deteriorating relations with the U.S., and the development of transnational clinical trials.

Managing the future is a central feature of Chinese governance, where the state views the future as an evolving project of modernization to be anticipated and realized (Braester 2016; Zee 2017; Mason 2021). However, imagining the future elsewhere reveals an alternative possibility—when China fails to fulfill the future for rare disease patients, they turn towards a foreign country that holds a political rivalry with their homeland. The future is materialized in transnational clinical trials. These biomedical hopes become tangible through the stages of clinical trials, where the accumulation of promising results and progression from one phase to the next in the U.S. generate both anticipation and anxiety among Chinese patients. These patients also strategically navigate their everyday caregiving decisions to optimize their chances for transnational treatment.

In this paper, I argue that imagining the future elsewhere is an ongoing care practice for rare disease patients, shaped by both biotechnology and geopolitical dynamics. The stories of these patients underscore the dangers of rising nationalism and protectionism, highlighting the critical need for global collaboration in addressing healthcare challenges.

Endless pain, hope that is not understood, eternal mental strength -Ethnography of migrant family with young adult patient of sickle cell disease in France

Meng Kou

Sickle-cell anaemia is a rare hereditary blood disease in the French public health system. Due to its post-colonial background, the treatment of this tropical disease has multiple intersections of race, gender, economic inequality, history, and biomedical domination.

How do families of immigrants from sub-Saharan Africa cope with sickle cell disease in the decades following the birth of a child with the disease, and how do they go on to survive and thrive? This is particularly the case in the context of a public healthcare crisis, where adult patients are neglected in emergencies and the medical strategies required change as the individual patient moves from childhood to adulthood, with a transformation of the interactions with institutions such as hospitals, schools and work.

This ethnographic study involves participatory observation in public hospitals, patient associations and digital patient groups, as well as follow-up visits at home, starting in 2020. The researcher will analyse the trajectory of medical treatment of two migrant families, visiting 10-12 times between 2022 and 2024, to find out how the family's medical

strategy is implemented when a rare disease patient transitions from childhood to adulthood, and the interactions between parents, patients and medical experts.
Genetic therapy is the hope for a cure for sickle cell disease patients facing chronic pain and drug side effects. These patients are aware of the risks and seek opportunities for a cure. The challenge of returning to normal life and work after genetic therapy is huge.
Patients group also creates a sense of solidarity.

"Complicated" Veins in "Complicated" Women: Pelvic Venous Insufficiency as Condition Multiple

Zuzanna Kuffel

Historically, in medical research as women were neglected compared to men, veins were neglected compared to arteries. Veins are much more complicated and diverse than arteries as female bodies are more complex than male ones. For these reasons, pelvic venous insufficiency, a common, yet under-recognized cause of chronic pelvic pain and several other complaints in women, poses a significant challenge to patients and physicians alike, fitting into broader discussions about care in "emerging illnesses". Based on two months of fieldwork across which I conducted interviews with patients and doctors in Poland, in this paper, I present pelvic venous insufficiency as condition multiple. I unpack different ways in which pelvic venous insufficiency is a condition multiple and analyze the consequences of the intersection of the complexities of female bodies, veins, and the Polish healthcare. The multiplicity of pelvic venous insufficiency occurs on several levels such as anatomy, etiology, symptoms, diagnostic pathways, impact on relationships, and approaches to diagnosis and treatment. Even though officially it is not a rare disease, due to gaps in medical training it is still not acknowledged by many doctors. While some interventional radiologists and other specialists acknowledge it and treat it, there are no settled protocols for diagnosis and treatment guidelines. In this paper, I also uncover that forming an online epistemic community around this illness, patients make each other aware of the multiplicity of approaches to the management of their condition, which can be unsettling, especially when doctors also become part of this community.

Generatively Unsettling Care: On Distrust and Failure in Transition to Imagine New Forms of Care

Ilaria Lesmo

The healthcare transition for paediatric patients with chronic and complex conditions has become an increasingly relevant topic in healthcare policy in recent years. In the Piedmont region (Italy), specific initiatives have been launched (Lala, 2021; Lesmo and Bignamini, 2023, Nave and Bignamini, 2018): they aim to foster stronger collaboration among institutions, forms of knowledge, and subjects and to reinforce a patient-centred model of care. These projects also seek to empower patients' competences by strengthening the social networks surrounding them.
However, subjective experiences may diverge significantly from these institutional models. This contribution focuses on the experiences of parents of children with severe

chronic and/or rare conditions, whose narratives challenge the above-mentioned representation of good “transitional care”. As their children’s conditions worsened dramatically during adolescence, these families were compelled to reshape care practices, often on their own. Faced with inadequate healthcare support and limited economic assistance, parents had to figured out new relational and financial ways to provide care at home or within external institutions.
Throughout these transformations, medical specialists were not always available, information on prescribed therapies was often lacking, and the knowledge of parents and their families was frequently dismissed. In their narratives, parents consistently describe their experiences in terms such as “distrust”, “loneliness”, “desperation”, and “failure”. Nevertheless, as Murphy (2015) argues, experiences of violence and discomfort in matters of care may be “generatively unsettling” (Murphy, 2015). The parents’ accounts reveal alternative relational, economical, socio-cultural, and epistemological perspectives, which emerge as new frameworks for imagining and enacting care in transition.

Diagnosed and Dumped: hEDS Care and the Diagnostic Moment

Claire Mavis

In chronic and rare disease discourse, diagnosis is often framed as the long-awaited conclusion of a “diagnostic odyssey.” Yet for patients with hypermobile Ehlers-Danlos Syndrome (hEDS), diagnosis frequently marks a moment of rupture rather than of final resolution. Drawing on four months of ethnographic research with hEDS patients, this paper introduces the concept of ‘diagnosed and dumped’ to help describe the moment in which many patients receive a diagnosis but are then offered little to no follow-up care, treatment, or clinical guidance. Rather than helping to initiating a clear path for clinical care, the diagnostic moment often functions as an end to medical attention, leaving patients in a liminal space of self-management, ongoing uncertainty, and reliance on peer-led communities (Turner 1994). This transition, from naming to the ensuing lack of care, complicates traditional expectations of biomedicine (diagnosis to treatment to cure) and reveals a disconnect between clinical goals and patients’ lived experiences of chronic conditions (Bologh 1982; Jutel 2019). Patients, then, must reconfigure what care means, where it takes place, and who provides it, often turning to expansive online support communities in the wake of this gap (Ashtari 2022). Ultimately, this paper argues for a redefinition of diagnosis as a relational and ongoing process. A process that extends beyond naming and transitions into practices of collaborative care. In doing so, it aims to highlight the complex dimensions of post-diagnostic life and calls attention to how care is both unsettled and reclaimed in the context of chronic and rare conditions.

The Loop in Transition: Attending to Hormonal Intra-actions in Automated Diabetes Care

Sabina Vassileva

The paper explores how people living with type 1 diabetes navigate practices of care as new algorithmic technologies mediate the management of chronic illness. Hybrid closed-

loop systems—often called “artificial pancreas”—combine continuous glucose monitoring sensors, insulin pumps, and self-learning algorithms, and promise a transition in diabetes care toward automation and reduced burden by distributing control in the loop. Yet, these systems often fail to accommodate cyclical hormonal fluctuations associated with the menstrual cycle or menopause. Even though clinical evidence acknowledges the effects of elevated estrogen levels on insulin resistance and higher glycemia (Brunerová, et al. 2023), and increasing numbers of clinical trials include menstrual cycle as a variable (Levy, et al., 2022; Elhenawy, et al., 2024; Mesa, et al., 2024), these hormonal intra-actions remain sidelined both in clinical practice and algorithmic design.

Drawing on ongoing ethnographic research with users of both commercial and DIY loop systems, I examine how people attend to hormonal variability and work around the limits of algorithmic management: adjusting insulin profiles, overriding algorithmic suggestions, temporarily suspending automation, or reconfiguring settings in anticipation of hormonal shifts. These everyday practices not only highlight the persistence of human labor within algorithmic care, but also show how care transitions towards datafication require negotiations between the algorithmic and embodied knowledge. How is the relationship between diabetes and hormonal variability enacted despite not being accounted for by the loop? And how is the intimate attention to changing bodily needs transformed through communication with self-learning algorithms in relation to largely unpredictable hormonal intra-actions?

The Social Body in Focus: Constructing Personhood through Intimacy

Syeda Shajeela Shawkat

This paper explores how individuals with spinal cord injuries (SCI) in Bangalore, India, navigate long-term transitions in intimacy, care, and personhood in the aftermath of injury. Drawing on twelve months of ethnographic fieldwork at a rehabilitation center, in private homes, and through digital platforms, it examines how injury not only transforms physical capacities but also reshapes social relationships, emotional attachments, and the terms of recognition.

I argue that intimacy and care are overlapping, emotionally complex, and often contradictory processes through which personhood is reconstructed after injury. Rather than discrete domains, care and intimacy blur into one another—manifesting through altered touch, emotional withholding, reconfigured kinship, and the search for relational continuity amid bodily and social precarity.

This chapter shows how intimacy is never merely private. It becomes a terrain through which gendered expectations, normative timelines of adulthood, and the legitimacy of disabled futures are contested and negotiated. Touch emerges not only as a sensory act, but as a social medium through which presence, desire, and recognition are communicated—or withheld.

By foregrounding the sensorium of care and the fragility of intimate recognition, this paper contributes to anthropological conversations on chronicity, gender, and the social life of disability It does so by tracing how care and intimacy are continually unsettled—improvised, reconfigured, or withheld—amid long-term transitions in bodily, social, and affective life. As part of this multimodal approach, the presentation will include excerpts

from a graphic novel created based on the same fieldwork, offering a visual perspective on intimacy and care after SCI.

In limbo. Entanglements of care, ethics, and law in transition processes of people with rare diseases

Malgorzata Rajtar

Like in other chronic and rare conditions, when people with rare metabolic diseases reach adulthood, they are no longer entitled to the care of their pediatric healthcare team. While their genetic condition does not change, their legal status does and their access to healthcare services deteriorates. At the age of 18, they suddenly become autonomous citizens, charged with managing their own care, irrespective of their emotional, intellectual, and practical capability to do so. Their current care givers, including their parents, can no longer represent them in healthcare contexts.

This paper draws from an ongoing ethnographic project and preliminary survey research on transition experiences of adults with rare metabolic disorders and their caregivers in Poland. While providing the wider context of this messy transition process, I focus on "legal incapacity" as an ethical and legal notion, which parents of adults with rare metabolic diseases and intellectual disabilities are faced with to ensure the continuity of care. Attending to transition, I illuminate how legally prescribed estrangement creeps into a dividual relationship characteristic of parent-patient units in rare metabolic diseases (Rajtar 2025). I further examine how legal and structural solutions inhibit providing good care and engender ethical challenges. In doing so, I intend to expand anthropological approaches to "unsettled" care (Cook and Trundle 2020).