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Advances in the Interdisciplinary Care of Children with Klinefelter Syndrome

Shanlee Davis, MD[a,b], Susan Howell, MS, CGC, MBA[a,c], Rebecca Wilson, PsyD[c], Tanea Tanda, BS[a,c], Judy Ross, MD[d,e], Philip Zeitler, MD, PhD[a,b], Nicole Tartaglia, MD, MS[a,c,*]

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Keywords

• Klinefelter syndrome • 47,XXY • Sex chromosome aneuploidy

Key points

- Klinefelter syndrome is a common but underdiagnosed genetic condition with significant phenotypic variability in childhood.
- The pediatrician needs to be aware of the increased risk for neurodevelopmental, psychological, and medical conditions that are associated with an additional X-chromosome.
- Over the next decade, we anticipate a sharp increase in diagnosis rates with advances in genetics, particularly prenatal and neonatal diagnoses.

Klinefelter syndrome (KS) is a common genetic disorder characterized by an additional X-chromosome in male individuals leading to a karyotype of 47,XXY. The clinical syndrome was first described nearly 75 years ago in several male individuals with small testes, tall stature, gynecomastia, and azoospermia [1]. Our construct of what KS entails has greatly changed

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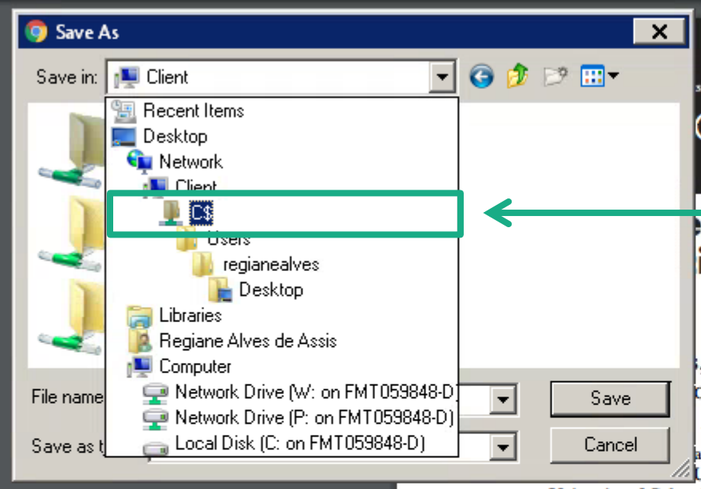

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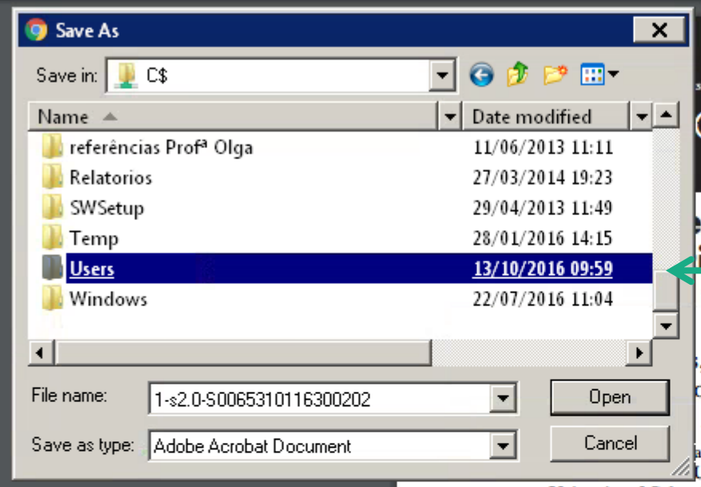

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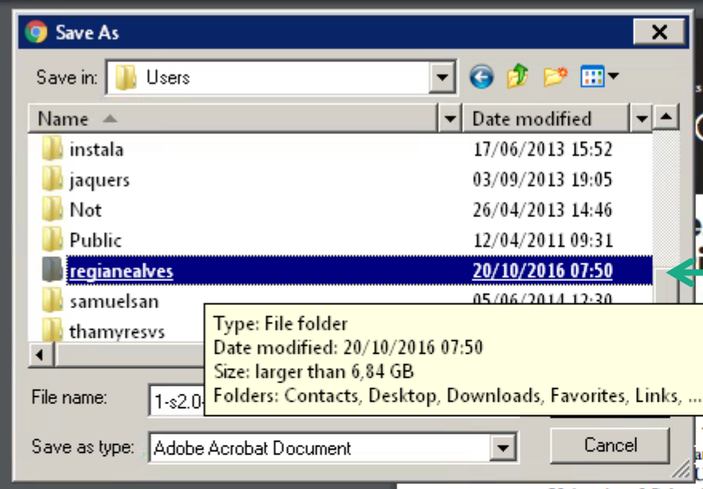

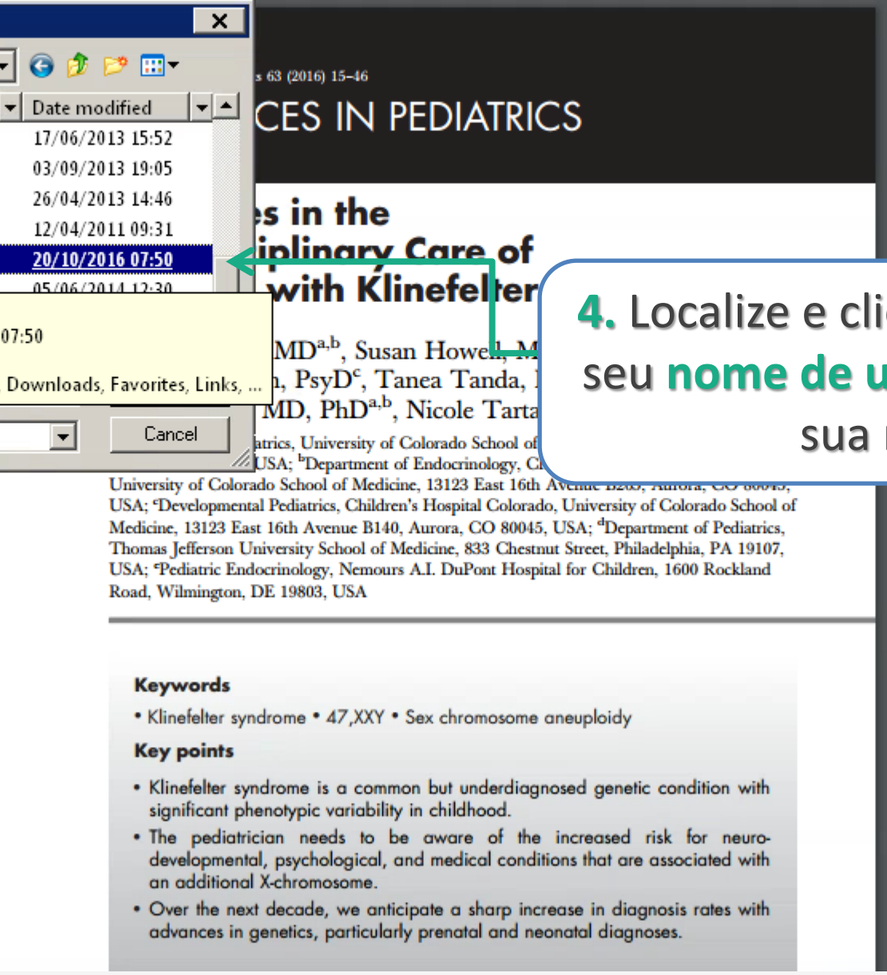

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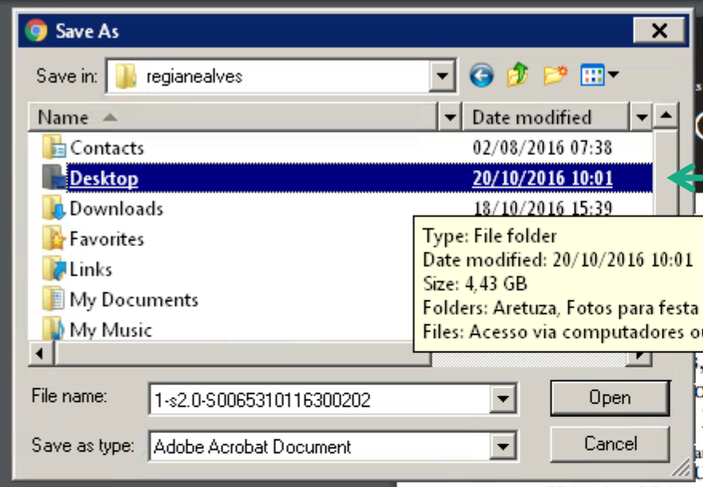

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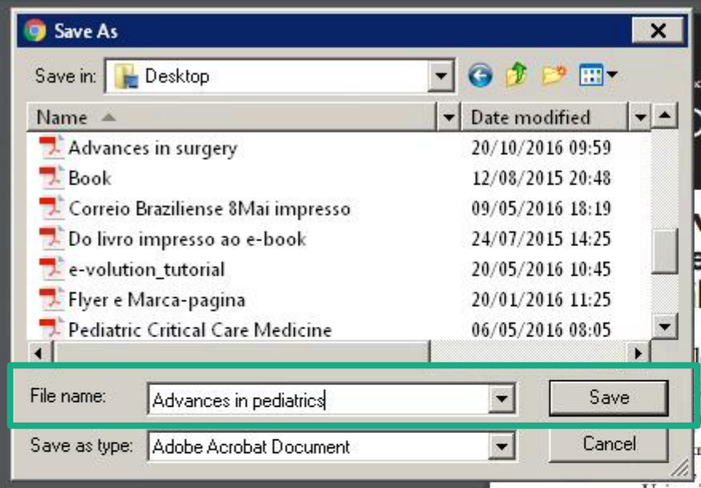

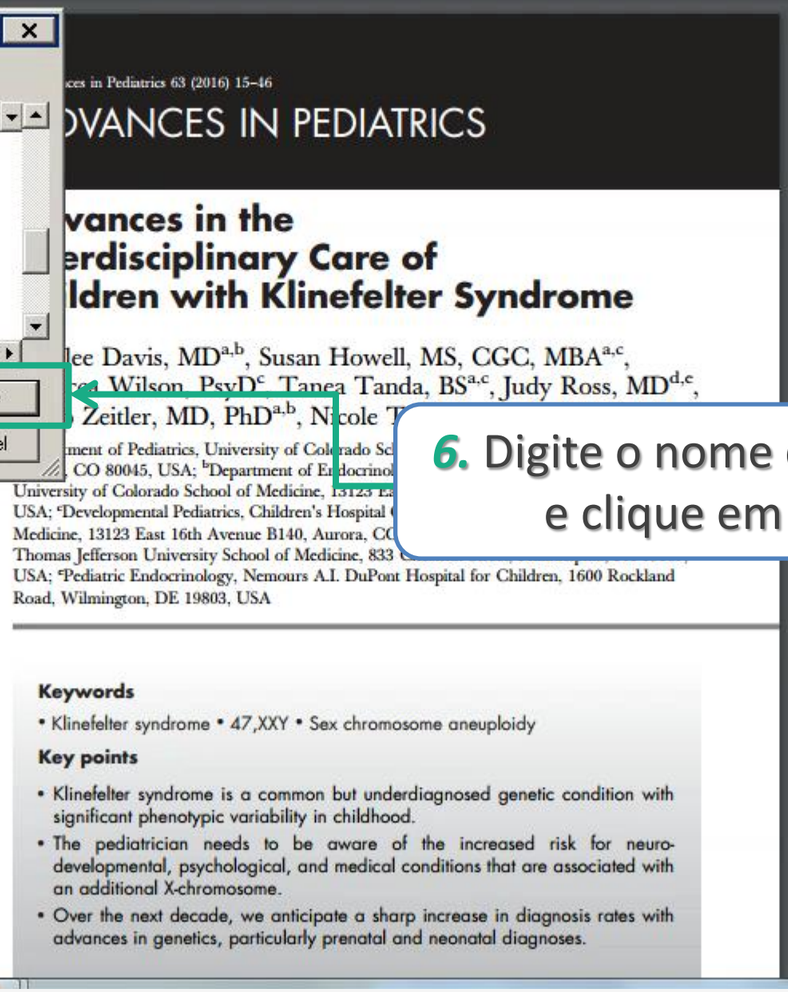
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8 Haakenstad A, Angelino O, Irvine CMS, et al. Measuring contraceptive method mix, prevalence, and demand satisfied by age and marital status in 204 countries and territories, 1970–2019: a systematic analysis for the Global Burden of Disease Study 2019. *Lancet* 2022; **400:** 295–327.

9 United Nations Department of Economic and Social Affairs. Transforming our world: the 2030 agenda for sustainable development. https://sdgs.un.org/2030agenda (accessed June 11, 2022).

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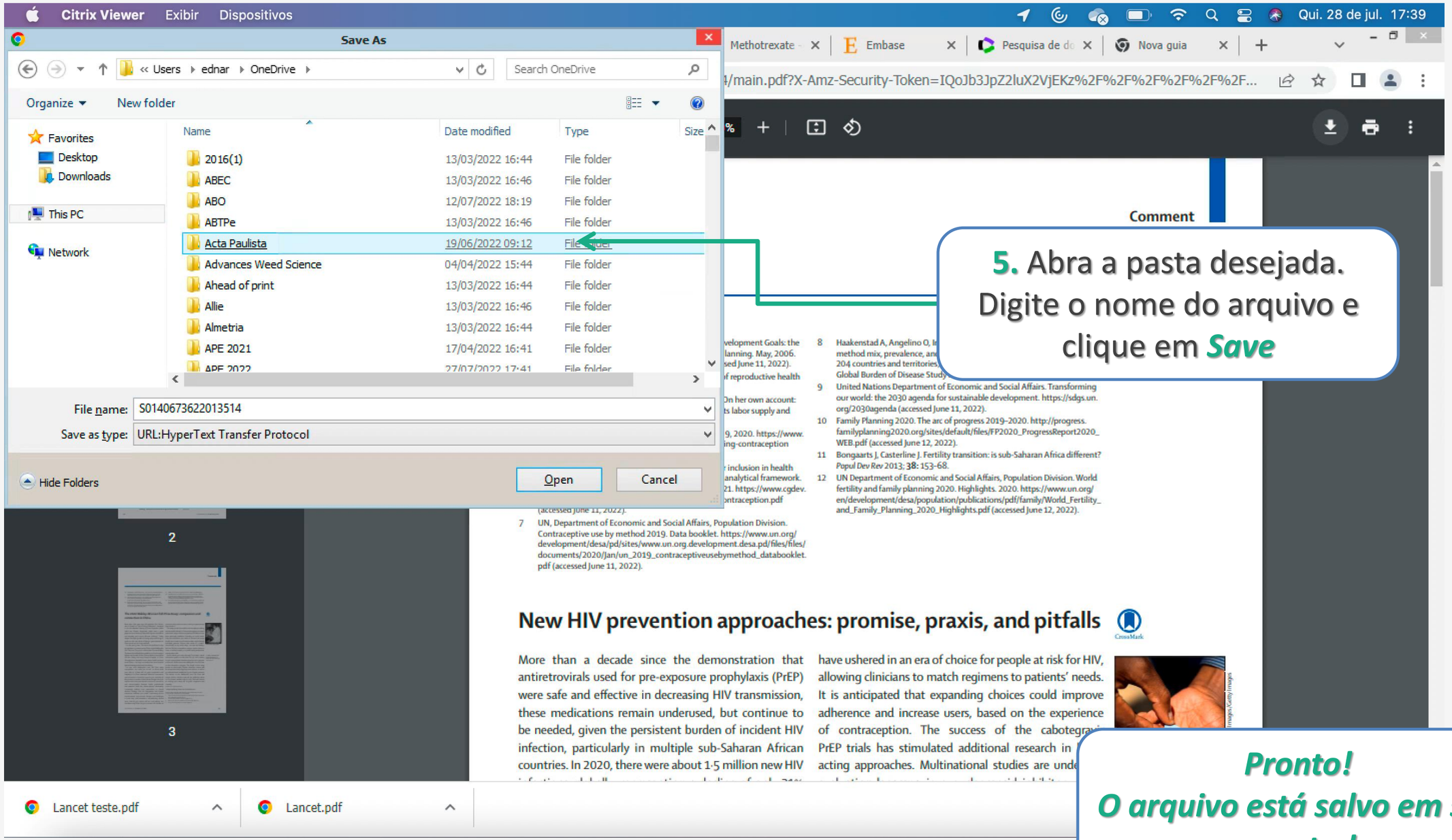
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