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CfDNA NGS-liquid biopsy: a new revolutionary approach for Klippel-Trenaunay Syndrome

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Klippel-Trenaunay Syndrome

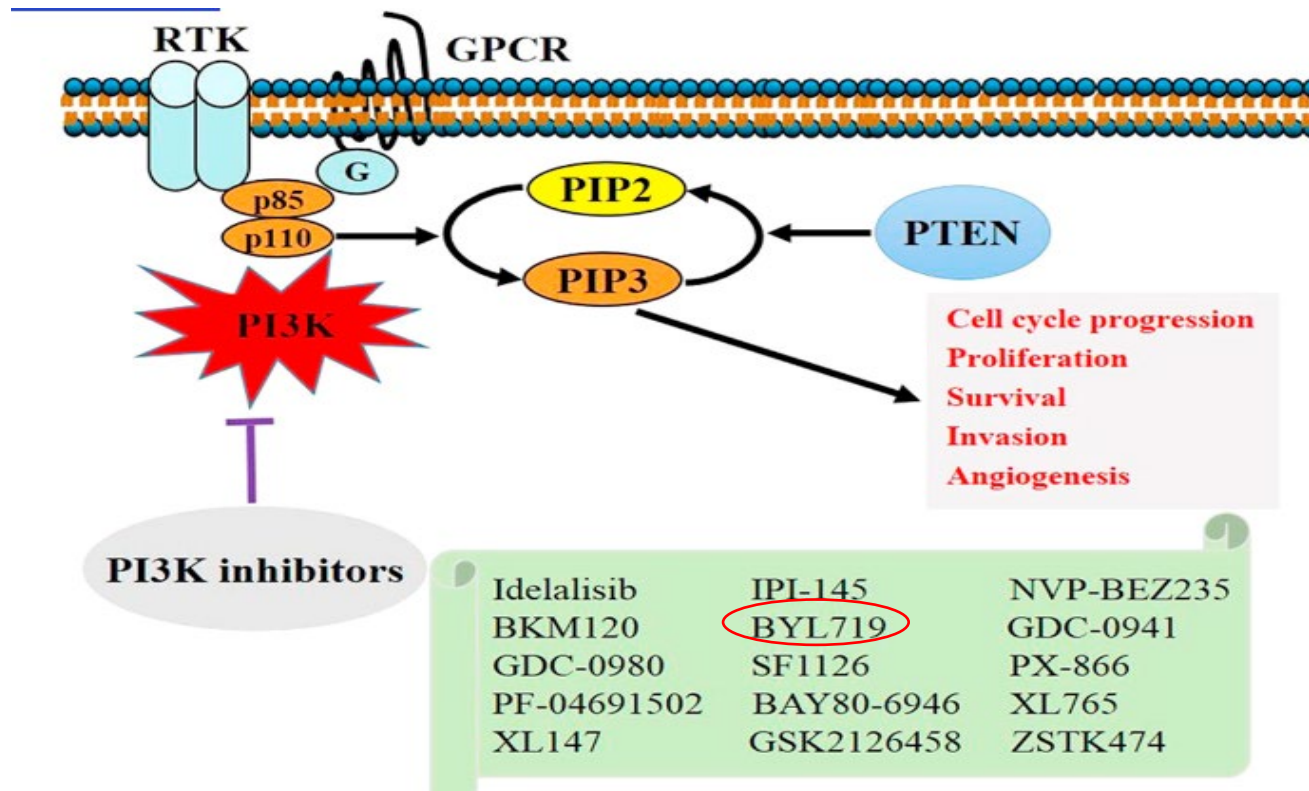
PIK3CA
SOMATIC MUTATION

Targeted therapy in patients with PIK3CA-related overgrowth syndrome

Quitterie Venot, Thomas Blanc, +33 authors · Guillaume Canaud · Published in Nature 2018 ·



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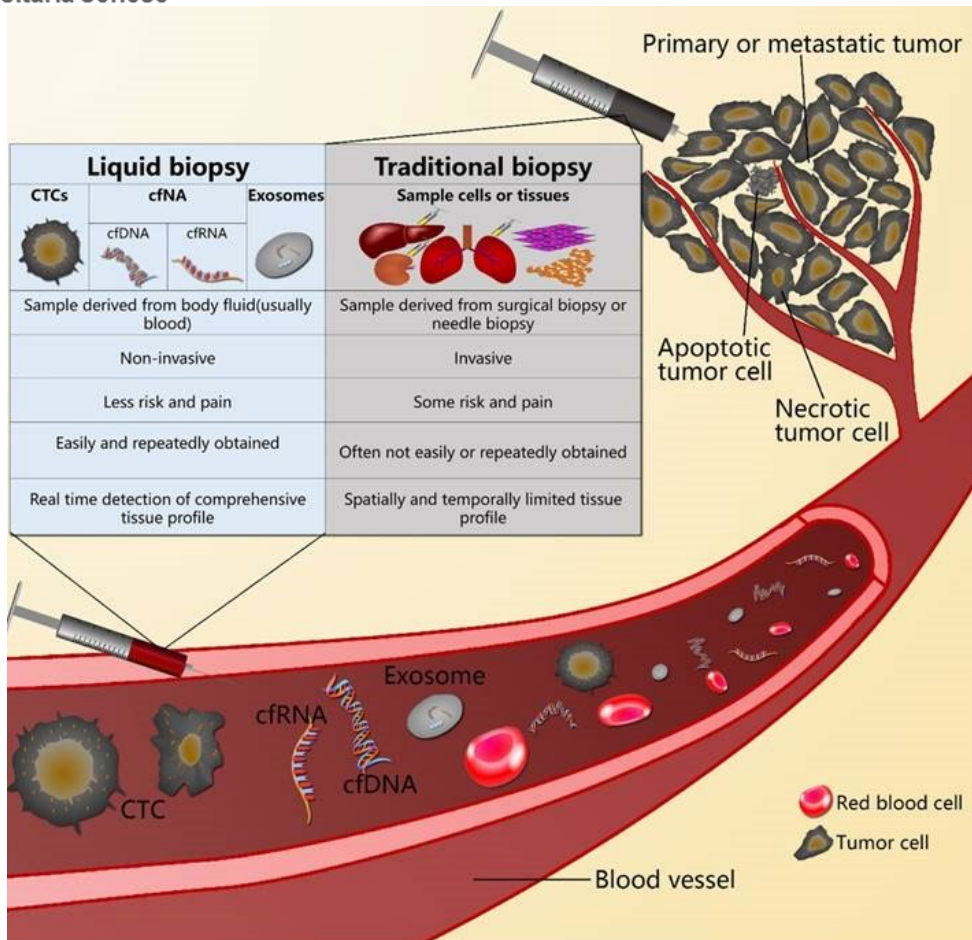
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CfDNA NGS-liquid biopsy

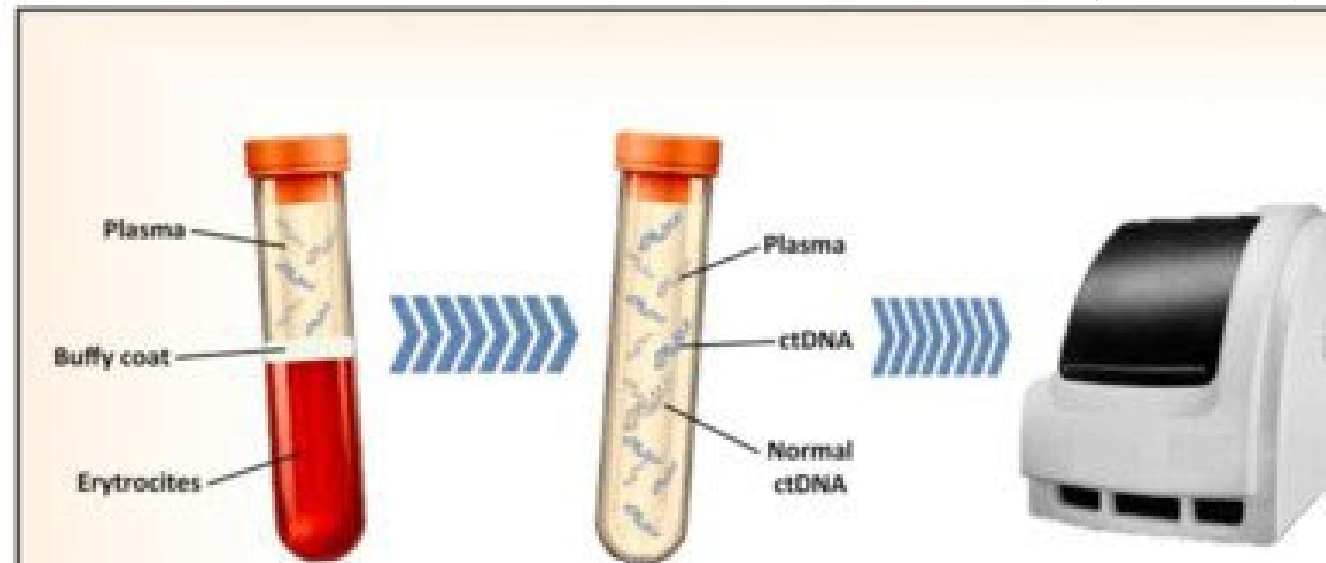


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Cell-free DNA



Next Generation Sequencing





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PIK3CA cfDNA-NGS Liquid biopsy results

Case	Age	KTS Phenotype	Limb hypertrophy	Vascular nevi/ Port-wine stain	Varicose vein	Mutations	Peripheral NGS liquid biopsy	Efferent vein NGS liquid biopsy
1	55	monolateral	dx leg	no	Yes	E545G	0,23%	na
2	34	monolateral; pancreatic cystadenoma	dx leg	Port-wine stain	Yes	E542K	0,21%	0,75%
3	45	bilateral	both legs	Vascular nevi	Yes	na	na	0,18%
4	61	monolateral with dx foot macroductyly	dx leg	Vascular nevi	Yes	E453K	0,36%	na
5	58	monolateral	sx leg	Vascular nevi	Yes	na	na	1,09%
6	26	monolateral	dx leg	Vascular nevi	Yes	M1043I	1,47%	na
7	38	monolateral	sx leg	no	Yes	E545K	0,96%	1,23%





Conclusions



- ❖ CfDNA NGS-Liquid biopsy from the efferent vein as a new propose to replace the tissue biopsy in order to detect somatic *PIK3CA* mutations
- ❖ The non-invasiveness of the method avoids any risk of bleeding
- ❖ The greater ease of the diagnostic technique increases the possibilities of personalized treatment tailored to the genetic defect





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European
Reference
Network

for rare or low prevalence
complex diseases

Network
Adult Cancers
(ERN EURACAN)

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Thank you...



Medical Genetic of Siena
Department of excellence 2018!
Director: Alessandra Renieri



Dr. Massimo Vaghi
Vascular Surgeon specialist
Ospedale Maggiore di
Crema



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