SURVEILLANCE PROTOCOL IN CARRIERS OF GERMLINE DISEASE-CAUSING TP53 VARIANTS

Exam	Periodicity	Age to start	Age to end	Condition	Evidence*
Clinical examination with, in children, specific attention to signs of virilisation or early puberty and measurement of blood pressure and, in patients who received radiotherapy, to occurrence of basal cell carcinomas within the radiotherapy field	Every 6 months	Birth	17 years		Moderate
	Annual	18 years	-		Moderate
Whole-Body MRI without gadolinium enhancement	Annual	Birth	-	High cancer risk TP53 variant** or patient previously treated by chemotherapy or radiotherapy	Moderate
		18 years	-		Strong
Breast MRI	Annual	20 years	65 years		Strong
Brain MRI***	Annual	Birth	18 years	High cancer risk TP53 variant	Moderate
		18 years	50 years		Moderate
Abdominal ultrasound	Every 6 months	Birth	18 years		Strong
Urine steroids	Every 6 months	Birth	18 years	When abdominal ultrasound does not allow a proper imaging of the adrenal glands	Weak
Colonoscopy***	Every 5 years	18 years	-	Only if the carrier received abdominal radiotherapy for the treatment of a previous cancer or if there is a familial history of colorectal tumours suggestive of an increased genetic risk	Weak

*This grading is based on published articles and expert consensus.

**A germline disease-causing TP53 variant should be considered as "high risk" if the index case has developed a childhood cancer; or childhood cancers have been observed within the family; or this variant has already been detected in other families with childhood cancers; or this variant corresponds to a dominant-negative missense variant.

***The first scan should be conducted with I.V. Gadolinium enhancement; in children, brain MRI should alternate with the Whole-Body MRI, so that the brain is imaged at least every 6 months.

Surveillance protocol

Li-Fraumeni and Heritable TP53-Related Cancer (hTP53rc) syndromes

This guideline has been drawn from the best available evidence and the consensus of experts in this area and it is regularly updated to reflect changes in evidence.

The expectation is that clinicians will follow this guideline, unless there is a compelling clinical reason specific to an individual patient not to.

European Reference Network
for rare or low prevalence complex diseases

Network
Genetic Tumour Risk Syndromes (ERN GENTURIS)

www.genturis.eu