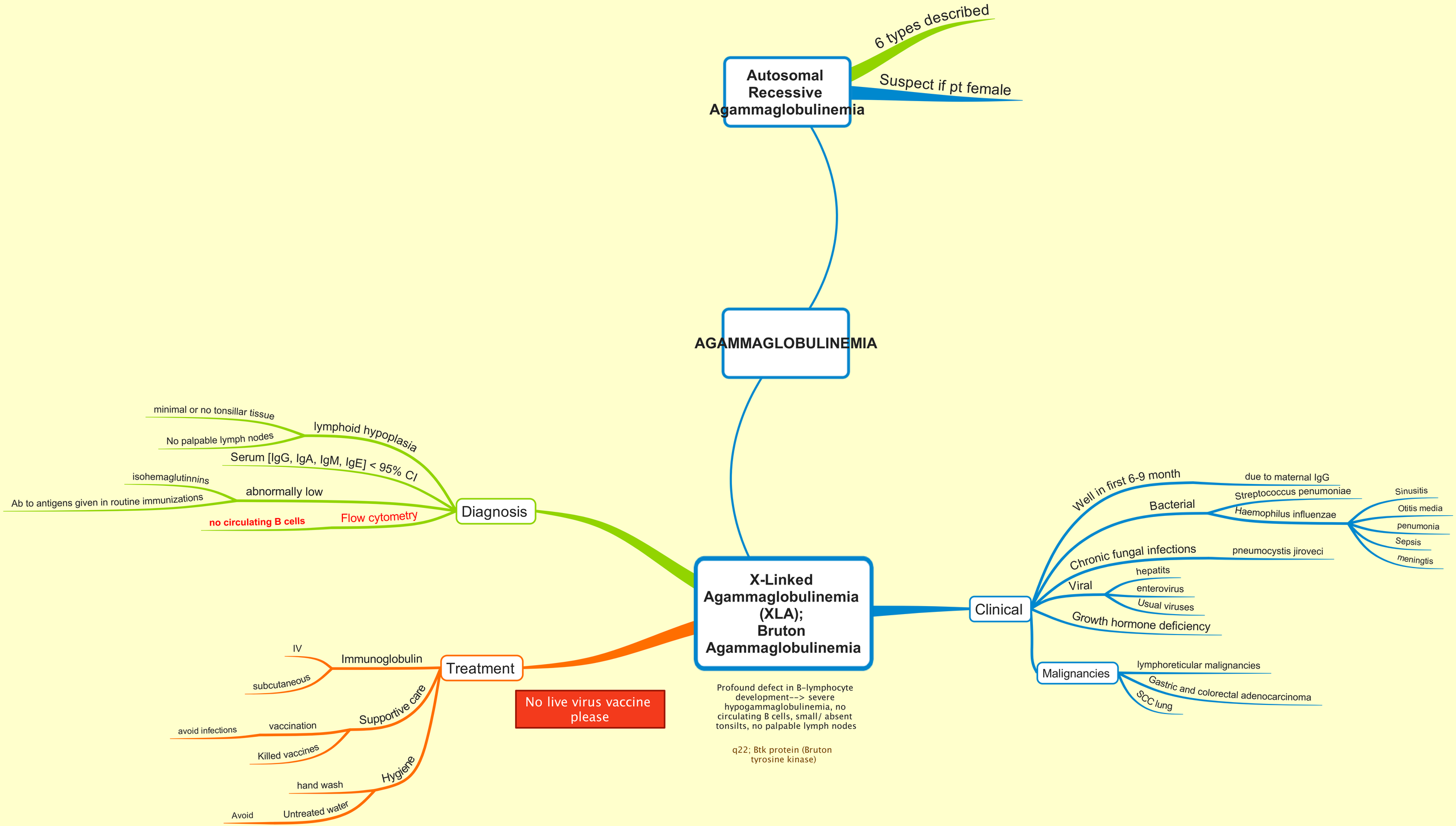
Autosomal Recessive Agammaglobulinemia
6 types described
Suspect if pt female
AGAMMAGLOBULINEMIA
X-Linked Agammaglobulinemia (XLA); Bruton Agammaglobulinemia
Profound defect in B-lymphocyte development--> severe hypogammaglobulinemia, no circulating B cells, small/ absent tonsilts, no palpable lymph nodes
q22; Btk protein (Bruton tyrosine kinase)
Diagnosis
lymphoid hypoplasia
minimal or no tonsillar tissue
No palpable lymph nodes
Serum [IgG, IgA, IgM, IgE] < 95% CI
abnormally low
isohemaglutinnins
Ab to antigens given in routine immunizations
Flow cytometry
no circulating B cells
Treatment
Immunoglobulin
IV
subcutaneous
Supportive care
vaccination
avoid infections
Killed vaccines
Hygiene
hand wash
Untreated water
Avoid
No live virus vaccine please
Clinical
Well in first 6-9 month
due to maternal IgG
Bacterial
Streptococcus penumoniae
Haemophilus influenzae
Sinusitis
Otitis media
penumonia
Sepsis
meningtis
Chronic fungal infections
pneumocystis jiroveci
Viral
hepatits
enterovirus
Usual viruses
Growth hormone deficiency
Malignancies
lymphoreticular malignancies
Gastric and colorectal adenocarcinoma
SCC lung